A TEXT BOOK
ON
ANIMAL
GENETICS

कृपया रेषा मारुन
पुस्तक खराब करु नये.

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Dedicated to
My Parents
and
Genetics Teachers

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PREFACE

Genetics is a fascinating subject of Biological Science which offers a lot of scope for the welfare of the mankind. During my teaching of the subject, I became aware of the need of a simple book which could promote the understanding of the principles of Genetics among the students.

This book is a complete introductory note containing divergent topics of Genetics under one cover. The subject has been presented in the sequences in which the discoveries in the Science of Genetics were made commencing with the Mendelian Genetics covering Cytogenetics, Molecular Genetics and Population Genetics culminating with the Applied Genetics. The text material is arranged in twelve parts and 40 chapters. Several examples and illustrations have been incorporated in all chapters. Many thought provoking questions and unsolved problems have been provided at the end of the chapters to enable the student to review the grasp of the subject and to serve as an aid for better understanding of the subject. The book also includes a comprehensive glossary on the subject.

Much of the contents of this book are the outcome of the number of discussions with my teachers, colleagues and students, and also of the consultation with the library.

This book is useful for all the science students especially for the veterinary science students. This book is written covering the Indian Veterinary Council Syllabus to make it applicable in the institutions following IVC syllabus.

I express my sincere thanks to the Vice-Chancellor, Tamil Nadu Veterinary and Animal Sciences University, Chennai for offering me this opportunity. My acknowledgement is due to my family members for all the help they rendered during preparation of this book.

I accept the responsibility for any errors and lapses and I hope to receive understanding pardon.

(P. KANAKARAJ)
Author

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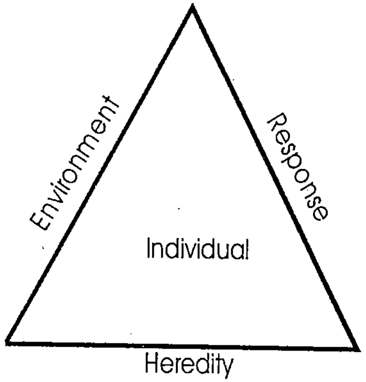
Chapter 1

Introduction

Genetics, is the branch of Science of Biology. It is one of the most fascinating subjects which deals with the mechanism of inheritance of traits and their transmission from one generation to the other. The word 'Genetics' was coined by William Bateson, an Englishman at the Conference of Plant Breeding in 1906. The word 'Genetics' is derived from the Greek root Gen, meaning to become or to grow into. Genetics concerns with the study of heredity and variation. Heredity in turn concerns with the mechanism of inheritance. Inheritance is carried on through the reproductive process, which involves transmission of Genes, the physical elements which are the units of heredity transmitted from parents to offspring.

Biological inheritance is defined by Prof.W.E. Castle as "Organic Resemblance based on descent". Like Be gets like. Chip of the same block are old sayings. Thus every species in this world reproduces only its own kind. However, all members of the species are not alike. It is said that even identical twins show variations in some characters. Variation thus is the law of nature. There are variations among the members of the same species with respect to observable characters like size, shape, weight,

Triangle of Life



Heredity - the primary factor.
Environment } the secondary factors
Response

colour, etc. Observations on plants and animals suggest that natural laws account for hereditary variations.

It was realised quite early in the study of Genetics that three factors act together to determine the characteristics of an individual. An individual is the result of an interaction of these factors namely heredity, environment and response. Heredity can be depicted as the base of the triangle. It is the primary factor. Environment and response are the essential secondary factors.

Of these, heredity is considered to be the primary factor in the study of Genetics since heredity determines what an individual is even before birth. It determines the characteristics of the future generation. Environment and response are the essential secondary factors. Environment is what the individual has such as food, shelter, and other favourable factors or obstacles to overcome. Response is what the individual does with the environment and heredity. All these three factors are tagged on to one another in determining the individual. Hence the study of Genetics helps to understand the principles of inheritance of characters and how these principles facilitate directional control on the mechanism of inheritance for the welfare of the mankind.

METHODS FOR THE STUDY OF GENETICS

There are at least six avenues by which the mechanism of heredity and variation, in the study of Genetics could be approached.

1. Observational method:

What is actually observed is taken into consideration. The cause of observed variation at successive generation from the original types, was more speculative rather than the actual principles underlying these consequences. Hence it became necessary to subject these observations to experimental procedures.

2. Experimental method:

Actual breeding experiments are conducted. These include twin studies, pedigree analysis, etc.

3. Biometrical methods or statistical methods:

Where actual breeding tests can not be resorted to, as in the case of human beings, statistical procedures are adopted. This was invented by Sir Francis Galton of Britain.

4. Cytological method:

The study of cell structures, particularly, the germ cell by microscopical examination of tissues which revealed the exact make up and behaviour of the genes. Such studies were initiated by E.B. Wilson and McClung.

5. Developmental method:

It is concerned with the ways in which the invisible hereditary genes emerge into visible characters.

6. Biochemical method:

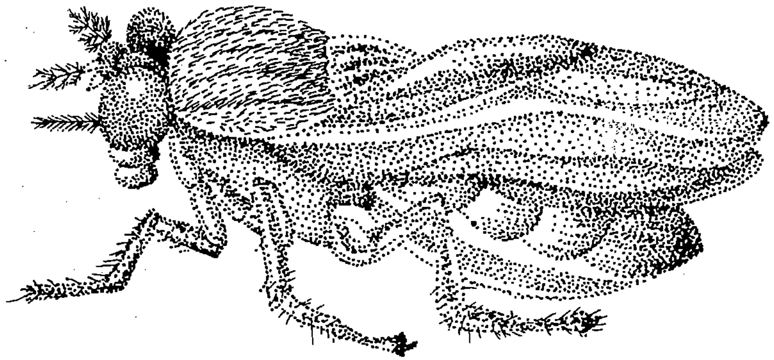
All inherited disorders will be understood in biochemical terms and the basic principles to determine many other inherited diseases will be found.

Advances are now being made in the biophysical aspects of Genetics that are designed to discover what the gene is and how it acts.

MATERIALS FOR THE STUDY OF GENETICS

An investigator, while choosing the material suited for experimentation for studies on basic mechanism, the organism of choice should have i) short life cycle ii) abundant progeny iii) a large amount of variability and iv) convenience in maintaining the material. In humans, for example, eye colour and number of fingers etc. can be investigated only by collecting the history of the families in which they occur, since ethics of human conduct and behaviour preclude the possibility of controlled experiments. Drosophila melanogaster, commonly known as the Fruit fly, fulfils the requirement of a good experimental material and is used extensively in genetic studies. A pair of flies can multiply to hundreds in a single generation of a short duration of only 10 days and in addition possesses a large number of variable characters like colour of eye, shape of the eye, body colour, length of the wing, wing venation, bristle number, etc. Mice, bacteria, hemospora (bread mould) and viruses also serve as good materials for study.

Drosophila Melanogaster



Drosophila melanogaster - fruit fly has short life cycle - 10 days more number of progenies, large number of variable characters

BRANCHES OF STUDY IN GENETICS

Depending on the kind of organism that is being studied, Genetics may branch off into Human Genetics, Plant Genetics, Animal Genetics, Drosophila Genetics, Microbial Genetics, Fungal Genetics, etc.

On the functional basis, Genetics may branch off into the following kinds:

1. Mendelian Genetics:

Study of the heredity of the clear-cut and contrasting characters like the eye colour, skin colour, feather pattern, normal vision or blind etc. known as 'Mendelian Traits' come under Mendelian Genetics. These traits are named after the scientist who discovered the mechanism of inheritance of such traits.

2. Quantitative Genetics:

Characteristics like height, weight, milk yield, egg production, wool yield, etc. which have to be measured are known as 'Metric Traits'. Biometry is a branch of Genetics which deals with Quantitative traits. Thus study of Quantitative traits is made with a population only. The study of heredity, when extended to a group of individuals or a population, it gets the name as 'Population Genetics'.

3. Cytogenetics:

A close study of the structure and functions of the cell, in which the genetic make up of an individual is coded in thread like structures, the chromosomes, with reference to heredity is known as cytogenetics.

4. Molecular Genetics:

It deals with the study of chemical structure of the hereditary material in relation to the control of cell activity. This has even led to the synthesis of the complex chemical molecule of which the hereditary material is made of.

5. Physiological Genetics:

It deals with the precise physiological pathways, which lead to the expression of the trait. Such kind of study has been termed as 'Physiological Genetics'.

6. Biochemical Genetics:

It deals with the biochemical basis of the inherited disorders.

7. Immunogenetics:

The general area of knowledge associated with the genetic basis of immunity is called as immunogenetics.

With more and more of specialisation, Nuclear Genetics, Radiation Genetics, Eugenics, etc. are in vogue now-a-days.

PRACTICAL APPLICATION OF GENETICS

The major use of Genetics are in the field of plant and animal breeding. Vast improvements have been achieved in poultry. From fowls, which laid only a few dozens of eggs in a year, new breeds and strains have been developed which lay 250-300 eggs in a year. The ancient cow which had just enough milk to feed the calf is now a mobile milk factory, because of judicious application of the principles of heredity in animal breeding.

The remarkable examples in plant improvement is the hybrid corn (maize) and new varieties of paddy whose yield is 50 per cent more over the best varieties of the previous years. New varieties of paddy, wheat, cotton have been developed to suit the new environment and give higher yields.

A knowledge of Human Genetics with respect to certain defects and diseases, will assist in giving the best treatment possible and also take prophylactic measures in time. Genetic counselling is the another field in Human Genetics which can help taking certain vital decisions in marriage, when the couple is confronted with certain hereditary defects and abnormalities like albinism, mental retardation, idiocy, etc. With the knowledge of Recombinant DNA, it is possible to detect viruses and other disease organisms in plants, animals and humans and to produce large quantities of a set of non-infective vaccines. An overall progressive advance in Genetics particularly in Molecular Genetics has opened new exciting possibilities for the welfare of the mankind.

EXERCISE

1. Define Genetics.
2. Who coined the word Genetics and what does it mean?
3. Explain the triangle of life.
4. Which is the suitable organism for Genetical studies and why?
5. Discuss on the different branches of Genetics.
6. Discuss on the application of Genetics for the welfare of mankind.

Chapter 2

History and Growth of Modern Genetics

Till the 17th century A.D. attempts to formulate explanation for inheritance was mainly based on speculations. Pythagoras, the Greek philosopher, proposed the theory that life comes from the moist vapour which descends from brain, nerves and other parts of the body of male. Aristotle, another Greek philosopher proposed a theory which is highly imaginative. He proposed that the female furnished the building material, while the male furnished the life giving power 'dynamics'.

Realm of speculation to reality was brought about during the latter part of the 17th century. The invention of microscope by Dutch Scientist Anton Van Leeuwenhock enabled observation of sperms in the semen. He also established the association of sperms with the eggs in frogs and fish and believed that sperm furnished the life of embryo, while the egg acts as a place of nourishment.

PREFORMATION THEORY

A short time later, another Dutch Scientist Jan Swammerdam developed the theory of Preformation. He believed that the development of embryo was only an enlargement of the parts that were already present in the sperm or egg. Regnier de Graaf (Dutch) studied the protubernances on the ovary and assumed them to be the eggs now known as Graafian follicles. He also suggested that the fusion of eggs and sperms contribute to the formation of embryo and disagreed with the preformation theory. Thus the nature of both egg and sperm as single cells and of fertilization as the union of their was not made clear until the latter half of the 19th century.

ORIGIN OF SPECIES

Various theories formulated so far had only helped to understand the nature of conception and formation of the embryo. Later, attention was focussed on the precise causes of heredity and variation leading to the origin of species and evolution.

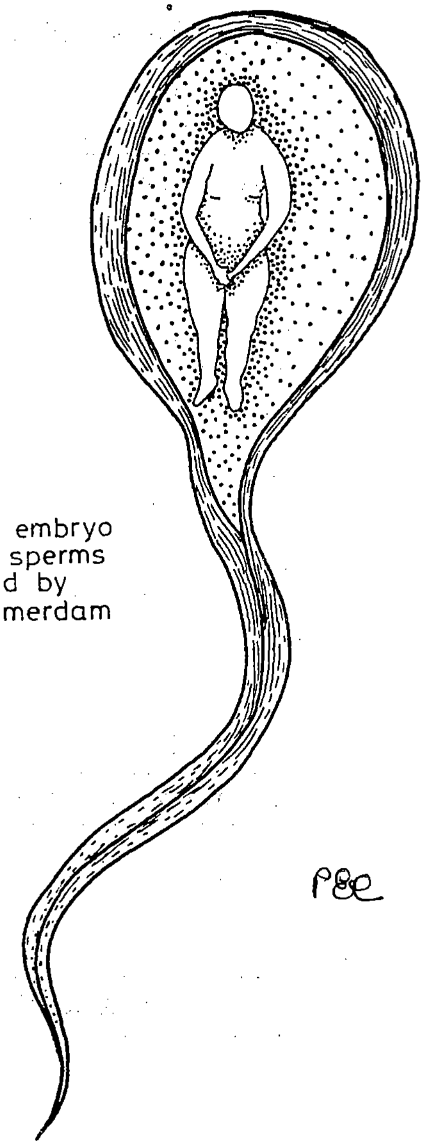
Carl Linnaeus (1707-1778), a Swedish Botanist evolved a system of classification, particularly in plants. He introduced the concept of species, in which he defined species as all individuals definitely marked by nature with certain characters. He believed that in animals or plants, characters are fixed by nature and those which did not conform to already existing species got destroyed.

THEORY OF LAMARCKISM

cael linnaeus

The cause for variation was best explained by Jean Baptist Lamarck (1744-1829), a French Scientist. The theory put forth by him in explaining the causes of variation is called "Lamarckism" which is quite significant in explaining the use and disuse of organs or body parts and inheritance of acquired characters. Any change in development of the use and disuse of the organs, due to necessity of adjustment to the environmental conditions during the life time of an organism, in due course, were transmitted to the offsprings. He proposed that an individual needs determined desire, its desire determine the use and disuse of its body parts, which brings about modifications of those parts and these modifications are in due course inherited. Heredity transmits the variations, thus acquired, leading to a new or different species. Environmental factors are thought to influence the response rather than the heredity.

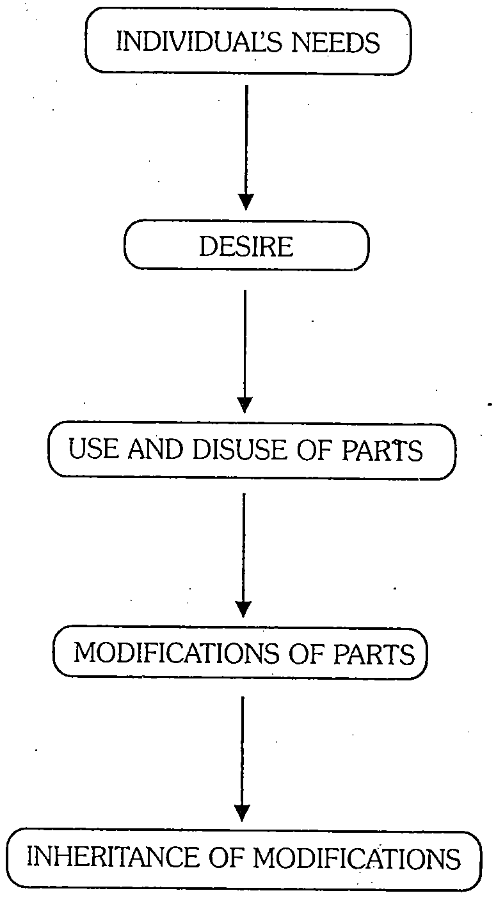
Preformation Theory



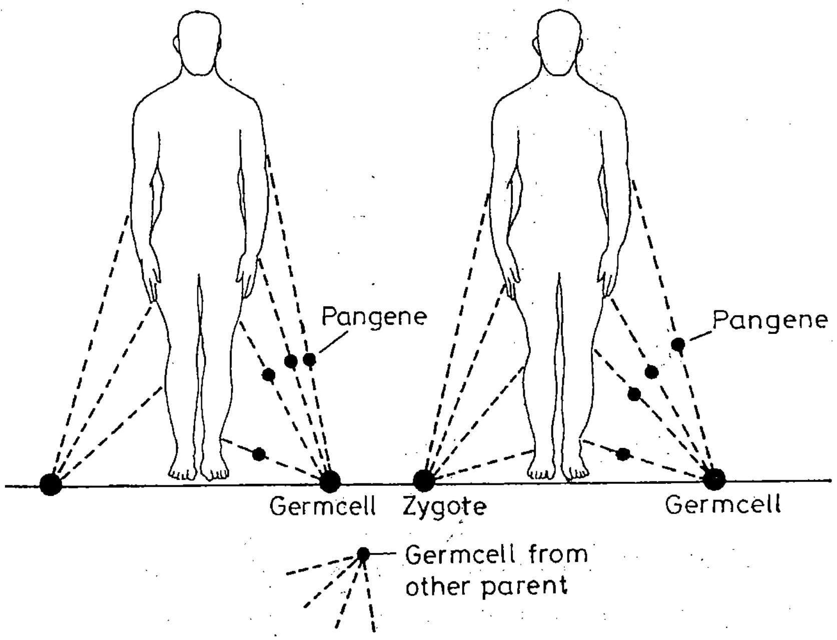
Miniature embryo
in human sperms
— reported by
Jan Swammerdam

Pee · cael

Theory of Lamarckism

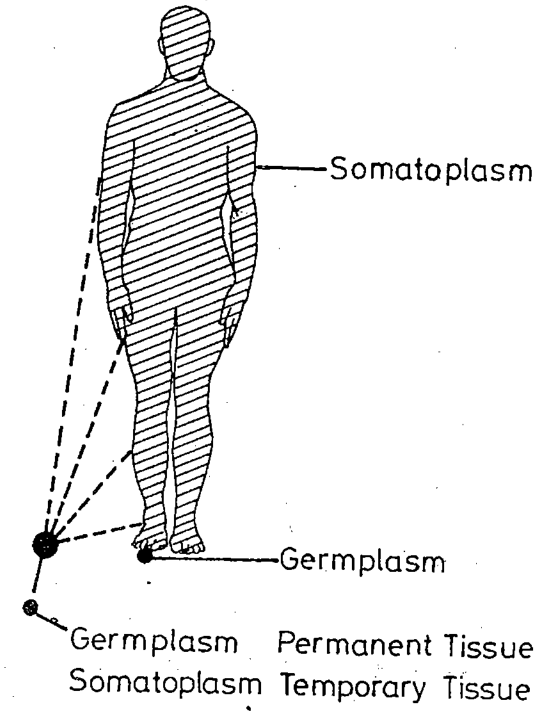

INDIVIDUAL'S NEEDS
DESIRE
USE AND DISUSE OF PARTS
MODIFICATIONS OF PARTS
INHERITANCE OF MODIFICATIONS

Theory of Pangenesis


Pangene
Pangene
Germcell
Zygote
Germcell
Germcell from other parent

Pangenes - Minute Particles present in all Cells

Theory of Germplams



THEORY OF EVOLUTION OR THEORY OF PANGENESIS

Charles Darwin (1809-1882) disagreed with Lamarck's views. According to Darwin, variation is an universally observable phenomenon in nature. Over production of offspring leads to a struggle for existence. In natural selection, only the fit survive through the elimination of the weaklings. Heredity continues the line of survivors with their successful qualities, so that eventually a new or different species may arise. As an explanation to heredity and variation, Darwin proposed the hypothesis of "Pangenesis". Every cell of the organs produced minute particles known as "Pangenes" which are carried through the blood, and reproductive cells also contain these pangenes. They conjoin to form the new cells and tissues resulting in a new individual, due to the blending qualities of both parents.

GERMPLASM THEORY

August Weismann (1834-1914), a German Biologist putforth the germplasm theory. Every metazoan organism is constituted of somatoplasm which makes up the body tissue and germplasm found in the germinal tissue, restricted to gonads. According to Weismann, the causes of variation are intrinsic i.e. within the germplasm and that germplasm is immortal. Somatoplasm is a transient tissue and germplasm is a permanent tissue, passed on from one generation to the other and not influenced by environment. A new individual is produced by the mingling of the germplasm of two parents.

MUTATION THEORY

In 1902, Hugo DeVries, a Dutchman, proposed another theory known as "Mutation Theory" for the origin of species. He observed among a group of evening primroses, the sudden appearance of new varieties which were dwarf with unusual leaf types, petals and other variants. He called this phenomenon as "Mutation" and the new varieties were found to breed true. He postulated that some of the old varieties suddenly gave rise to new ones, known as Mutation leading to the origin of new species.

MENDELISM

However, all these theories had the respective advantages and disadvantages in explaining the heredity and variation. It was Gregor Johann Mendel (1822-1884), an Austrian Monk who laid the foundation of Modern Genetics. He conducted his experiments on various plants, notably, Garden Peas (Peas Plant) which gave a lot of variations, with diversified characters. Mendel's experiment was simple and unique in that he chose one character at a time for his studies. He kept an accurate record of his observations which scored over his predecessors like Kolreuter. Mendel wrote his findings in 1865 and in 1866, and his results with Garden Peas appeared in the transactions of the Natural History Society at Brunn. However, Mendel's work did not gain appreciation during his life time. In 1900, three botanists, DeVries of Holland, Von Tschermark of Austria and Correns of Germany obtained identical results as published by Mendel earlier. Their rediscovery of Mendel's contribution was closely followed by several publications with the result of experiment on plants and animals, all confirming Mendel's earlier work. Entire work however, rightly came to be named after the original discoverer as Mendelism.

CELL THEORY

Second great principles of 19th century biology, the cell theory was putforth convincingly by the German Scientist Mathias Schleiden and Theodar Schwann, that all plants and animals are constructed from small fundamental units called cells and all cells arise from other cells, by the process of cell division.

GENE

Wilhelm Johannsen, ~~the~~ Danish botanist coined the word 'Gene'. A gene is a single unit of heredity. H.Nicolson Ehlc Swedish botanist made important discoveries about the nature of heredity in his work on wheat. Thomas Hunt Morgan, a Professor at Columbia was the first to conduct experiments on fruit fly Drosophila melanogaster. The present knowledge on genetics was built by many outstanding biological scientists.

EXERCISE

1. Explain on the following theories:
   a) Preformation theory → swammerdam.
   b) Theory of pangenes → Evolution → charles darwin
   c) Germplasm theory → August weismann .
   d) Theory of Lamarckism → Acquired Inheritance
   e) Mutation theory → Hugo . de vries .
2. Who is the father of modern genetics and write briefly about him?
   Gregor John mendel
3. Who coined the word `Gene'? – willam ~~Bateson~~
   Johannsen.

Chapter 3

The Physical Basis of Heredity

THE SMALLEST UNIT OF LIFE IS THE CELL

The physical connection between parents and offspring for transmission of hereditary capacities remains in the germ cells. The sperm and egg cells, like all other cells in the living organism, contain a nucleus, and it is the nucleus that must be concerned with inheritance.

The nuclei contain rod-shaped bodies called chromosomes. It is these bodies that constitute the material that transmits the determiners of characters and govern inheritance. The word chromosome being derived from the Greek word chromos (colour), and soma (body) is given to these bodies from the fact that they take certain stains more heavily than do other parts of the cell.

The cell theory was first promulgated by Schleiden and Schwann in 1839, affirming among other things that all organisms, both plant and animal are made up of cellular units and all cells arise from other cells.

A TYPICAL CELL

The bodies of all plants and animals consist of cells. That are diverse in size, shape, structure and function but are alike in their more fundamental characteristics. They are in general very small. Ordinary cells range from 0.1 to 0.01 mm in diameter, many of them are very much minute. Each typically consists of a denser mass of protoplasm, the nucleus, surrounded by cytoplasm.

THE NUCLEUS

The nucleus is the directive centre of most of the activities of the cell. The fact that male gametes in animals and plants ordinarily consist chiefly of a nucleus and yet equivalent to female gametes as carriers of inheritance indicates that this part of the cell is of major importance in the transmission of traits from generation to generation.

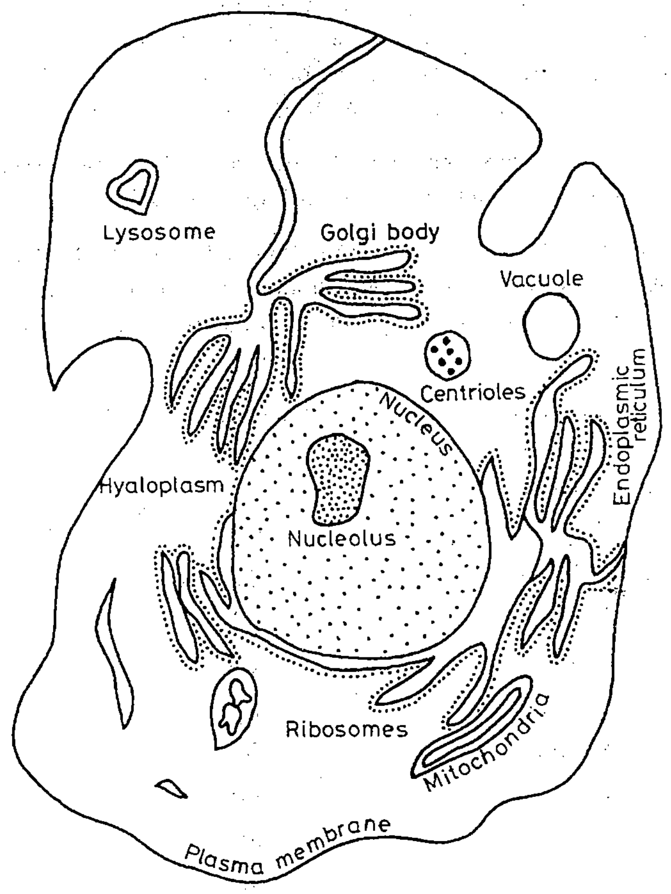
In cells which are not in the process of division, the nucleus is provided with a definite nuclear membrane. This surrounds a clear and homogenous nuclear sap in which is suspended a net work or reticulum of much denser substance, consisting of a material which stains very readily, the chromatin.

In addition to this reticulum most nuclei contain one or more dense, usually rounded bodies, the nucleoli. Most importantly, cells arise from cells by a process of cell division. Most cells are capable of growing and splitting roughly equally to give two daughter cells. At the same time, the nucleus divides so that each daughter cell can receive a nucleus.

THE CYTOPLASM

The protoplasmic material of the cell outside the nucleus is known as the cytoplasm. In most animal cells a series of fibrils, the Golgi apparatus is present. In animal cells there are also usually a definite centrosome or central body ordinarily surrounded by cytoplasmic rays, the aster. The central body seems to take a leading part in cell division. Certain granulated bodies known as mitochondria are present.

Animal Cell



Every cell has subcellular structures called organelles which many types of cells have in common. Most organelles are too small to be observed under light microscope but their structure can be studied under electron microscope. These organelles perform distinctive functions which in total produce the characteristics of life associated with the cell.

CHROMOSOMES: CHROMOSOME NUMBER

Any body cell other than sex cells (gametes) i.e. all the somatic cells contain one set of chromosomes inherited from the male parent (paternal) and one set of chromosomes inherited from the female parent (maternal). The number of chromosomes in this dual set is called the diploid (2n) number. The 'ploid' refers to chromosome sets.

Sex cells or gametes contain half the number of chromosome sets found in somatic cells, which are referred to as haploid cells (n).

The number of chromosomes in somatic cell is constant for the members of given species. For example human somatic cells contain 46 chromosomes, and the chromosome numbers of some species are given below:

Species	Chromosome numbers (2n)
Drosophila	8
Garden pea	14
Tobacco	48
Horse	64
Cattle	60
Buffalo	50
Sheep	54
Goat	60
Pig	38
Dog	78
Poultry	78

Functions of cellular organelles:

Cell organelles	Functions
Cell membrane	Differentially permeable membrane which allow selective substances to enter in and to liberate out.
Nucleus	Regulates growth and reproduction of the cell.
Chromosomes	Carriers of hereditary units — genes.
Nucleolus	May synthesise ribosomes.
Nucleoplasm	Contains materials for building DNA and messenger molecules which act as intermediates between nucleus and cytoplasm.
Nuclear membrane	Permits entry of selective substances from cytoplasm.
Cytoplasm	Contains machinery to carryout the instructions from nucleus.
Endoplasmic reticulum	Greatly expanded surface area for biochemical reactions which normally occur at or across membrane surfaces.
Ribosomes	Sites of protein synthesis.
Centrioles	Capable of replication— Form poles for the cell division
Mitochondria	Energy production.
Golgi body	Production of secretions.
Lysosome	Production of intracellular enzymes to eliminate the bacteria and other foreign substances.
Vacuoles	Storage depots for excess water, waste products, soluble pigments, etc.
Hyaloplasm	Contains enzymes for glycolysis and structural materials.

Morphology of chromosomes:

Each chromosome can be distinguished from others by the relative lengths of the chromosomes, the position of centromere, which divides the chromosomes into two arms of varying length, the presence of tiny terminal extensions of chromatin material called satellites. A chromosome with a median centromere is known as metacentric which will have arms of approximately equal length. A submetacentric or acrocentric chromosome has arms of distinctly unequal size. The chromosome which has its centromere at or very near one end of the chromosome is called as telocentric.

Autosomes - Sex chromosomes:

In mammals and Drosophila, males are heterogametic. Sex is associated with a morphologically dissimilar (heteromorphic) pair of chromosomes called sex chromosomes. They are labelled as x and y. Y chromosome determines maleness. Females have two morphologically identical x chromosomes (homogametic). All chromosomes other than sex chromosomes are called autosomes.

EXERCISE

1. Describe with diagram on the structure of Animal Cell.
2. Write the functions of important organelles in the Cell.
3. Give the chromosome numbers of Drosophila and domestic animals.
4. Give an account on morphology of chromosomes.

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Chapter 4

Cell Division

The overall reproductive cycle of a cell consists of doubling of all the components of the cell, followed by a division that distributes the components to the daughter cells. The essence of the plan is that the gametic material is packaged into a small number of chromosomes. In the period between cell divisions, in the interphase, the genetic material is contained within a nuclear envelope, but in a highly extended and attenuated form. The duration of interphase in plant and animal cells varies between 10 and 20 hours.

(During the period of division, which takes about an hour, the genetic apparatus goes through a complex but intelligible series of acts. The chromosomes condense to compact bodies. In most cases the nuclear envelope disintegrates. The chromosomes are now part of a mitotic apparatus. The mitotic apparatus is a large body, possessing definite poles, which represent the destination of the chromosomes, and its equator determining the plane through which the cell will divide.) The chromosomes are first moved to the equator. Then sister chromosomes, the products of the reproduction of each chromosome at an earlier time, split apart and move to the opposite poles. The cell now divides through the equator of the mitotic apparatus, producing two cells, each with a full set of replicas of the chromosomes that present cell received at the division in which it was born. The chromosomes of each daughter cell now uncoil. The new nuclear envelope is formed around them, and they are ready to begin careers that will end when each becomes two cells in the same way.

In plant and animal cells, actual replication of the genetic material - the doubling of DNA - takes place only between divisions. The replication of the chromosomes merely gives one cell with double set of chromosomes. To make two cells, these chromosomes must move into an equator defined by poles; then sister chromosomes move to opposite poles. One of the prerequisites of cell division, is the production of centrioles. The centrioles are generally found in pairs, and two centrioles commonly lie at right angles to each other. (However, centriole particles have not been seen in plant cells.)

A) MITOSIS

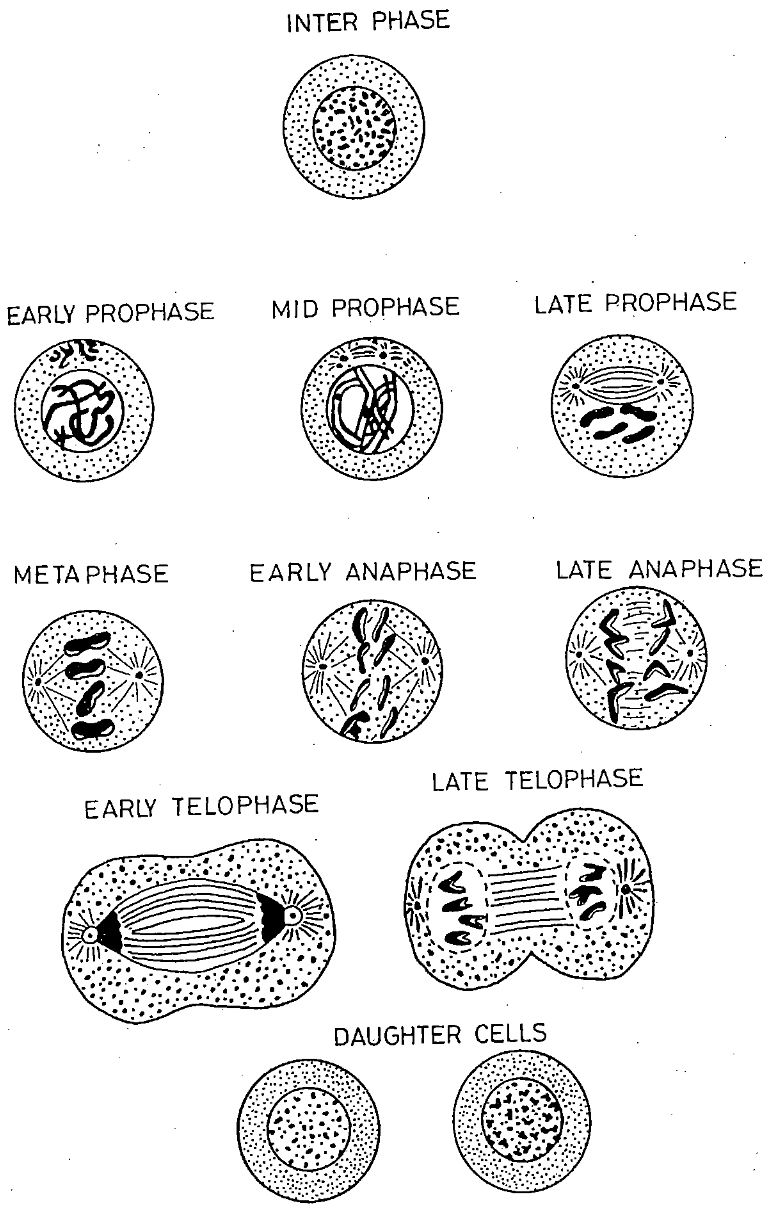
Each nucleus encloses a fixed number of linear bodies, the chromosomes. Before cell division, each chromosome duplicates to form two identical chromosomes, doubling the number of nuclear chromosomes. During nuclear division, one each pair of daughter chromosomes moves into each daughter nucleus. As a result of these events, the chromosomal complement of daughter cells is usually identical to that of parent cells.

This process by which two cells are made out of the one is termed mitosis. It occurs constantly, and particularly during growth, in all cellular organisms.

Events at mitosis:

1) Chromosomes are thin extended threads. Each of two parent centrioles is paired with a smaller centriole. At some point before division the chromosomes replicate.
2) The centrioles begin to separate and the spindle starts to form.
3) In prophase, the chromosomes coil, becoming highly condensed, the nuclear membrane and nucleus break down and the centrioles move apart to establish poles toward which the chromosomes move. At the

Mitosis



same time connections between centromeres and the poles are established.

4) In metaphase, chromosomes move to the cell equator, after splitting apart.

5) Sister chromosomes move toward poles during anaphase.

6) Telophase - the chromosomes uncoil, the nuclear membranes and nucleoli of the daughter cells are formed.

In mitosis, the particles of each chromosome attract similar ones to themselves during the resting stage, so that one thread reproduces or becomes two. This division is followed by the association throughout prophase of the two chromatids which arise from the division.

In general, although individual variations of the mitotic process and structures exist, the usual stages of mitosis can be described as follows:

Interphase: The interphase period between successive cell divisions consists of processes associated with growth and preparation for mitosis. In many cases, as compared to the period of active mitosis, which may range from about 10 minutes to a few hours, the interphase of "intermitotic" period of most cells is usually many times longer.

The resting cell is characterized by the presence of a nuclear membrane, a single centromere, and by a chromatin network within the nucleus.

Prophase: It is the first stage in which the preparations for cell division continue, but at a more active pace than during interphase. Chemical constituents of the new chromosomes are now synthesized in those cases where they have not been formed during interphase. Prophase includes the coiling of the chromosomes, or their condensation, to the point where they are now visible as thread like structures. As a rule, each of these mitotic prophase chromosomes appears longitudinally split into two duplicates (sister chromatids). Further prophase events are, the splitting of the centrosome and the movement of each half to opposite sides of the nucleus, the disappearance of the nucleolus, and the beginning of the breakdown of the nuclear membrane.

Metaphase: Each chromosome attaches through some point along its length, the centromere, to a double poled spindle shaped structures, the spindle. The spindle to which the centromeres attach is usually formed in animal cells through the separation of two cytoplasmic granules, called centrioles. As two centrioles separate, distinctive lines (astral rays) appear to radiate outward from each centriole, forming a network of continuous spindle fibres between them. Now chromosomes move and get arranged on a flat plane (metaphase plate) midway between the two poles of the spindle.

Anaphase: The shortest of the mitotic phases, occurs when the centromere of each pair of chromatids begins to function as a double structure. At that point, each centromere with its attached chromatid on the metaphase plate separates from its sister centromere and chromatid, moving toward opposite poles of the spindle. Thus, active repulsion between sister chromatids, each dragging its chromatid along with it occurs.

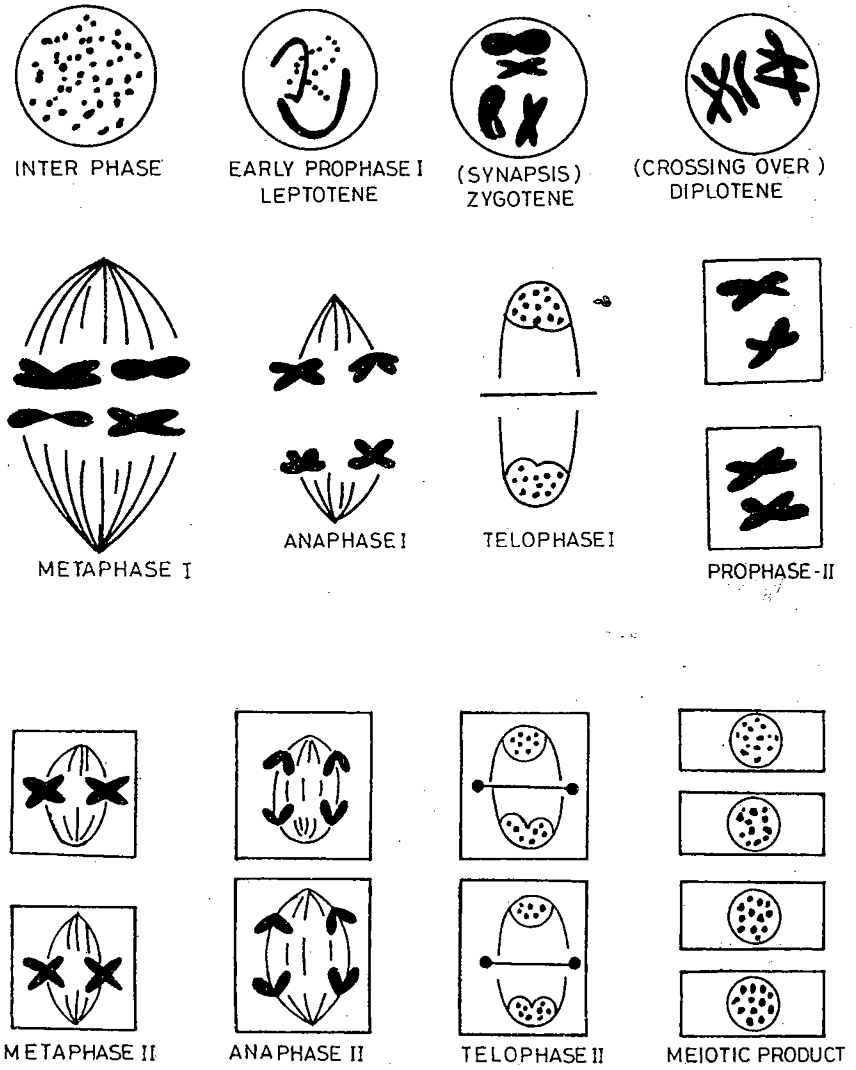
Telophase: In the beginning, the half-chromosomes grow until they attain full size and the division of the cell body into two parts becomes complete. The mantle fibres disappear, the nuclear membrane begins to reform around the chromosomes. At the end of telophase, the nuclear membrane becomes complete, the chromosomes break up into a chromatin network, and two resting cells take the place of the single one with which the process began.

B) MEIOSIS

Sexually reproducing organisms have a mechanism that enabled them to regularly reduce the number of chromosomes in each gamete to half, the usual number, diploid to haploid status. Meiosis is simply a process by which the chromosomes are separated in the sex cells and their numbers reduced from the diploid to the haploid condition.

The cell division which immediately precede the formation of sex cells, (Gametogenesis) occurs only in specialized cells of reproductive organs. Gametes contain the haploid (n) number of chromosomes, but originate from (2n) diploid cells of germ line. The number of chromosomes are reduced by half during gametogenesis. The reductional process is called meiosis. Meiosis actually involves two divisions. The first meiotic division

Meiosis



(Meiosis I) is a reductional division producing two haploid cells from a single diploid cell. The second meiotic division (Meiosis II) is an equational division which separates the sister chromatids of the haploid cells.

In this process each individual chromosome usually has a pairing mate or homologue. Upon fertilization, a diploid zygote is formed.

At the beginning of meiosis, the chromosomes appear as single threads and the separate homologues therefore attract one another. In the diploid, there are two homologues of each kind of chromosome. These chromosomes come to lie side by side in pairs, in a pairing process called synapsis.

First meiotic prophase:

As a rule, the meiotic divisions follow a standard scheme during which two successive divisions of chromosomes occur. One of these divisions represents a reduction division in which members of homologous pairs of chromosomes are separated into daughter cells without duplication i.e. their numbers are reduced to half. Thus initial pairing and subsequent separation of homologous chromosomes occur. This initial pairing occurs during the fairly lengthy first meiotic prophase, which can be subdivided into the following five stages:

Leptotene: The chromosomes first appear as long, slender threads, with many bead like structures (chromomeres) along their length. Meiotic prophase chromosomes generally appear as single and individual structures until the pachytene stage.

Zygotene: Homologous chromosomes appear to attract each other and enter into a very close zipper like pairing (synapsis).

Pachytene: A stage of progressive shortening and coiling of the chromosomes that occurs once zygotene pairing has been completed. At pachytene each homologous chromosome consists of two sister chromatids associated with two sister chromatids of their homologous partner. This group of four chromatids is known as bivalent or tetrad, and a series of exchanges of genetic material can occur or has already occurred between the homologous chromatids. These signify the genetic mechanism of crossing over or recombinations. A physical counterpart to genetic crossing over can be observed during two following prophase periods, diplotene and diakinesis.

Diplotene: At the diplotene stage each chromosome now acts as though they were repulsing their closely paired homologue. Distinctly visible separation now occur between homologous chromosomes except for specific regions where an actual physical crossing over appears to have taken place between homologous chromatids. These crossed areas, chiasmastar are x-shaped attachments between chromosomes and seem to be the only remaining force holding each bivalent together until metaphase.

Diakinesis: Coiling and contraction of the chromosomes continues until they are thick, heavy staining bodies. In this process, the bivalents usually migrate close to the nuclear membrane and become equally distributed. The nucleolus either disappears or detaches from its associated chromosome. During the latter part of this stage, or early part of metaphase, the nuclear membrane dissolves and the bivalents attach themselves by their centromere as to the rapidly formed spindle.

First metaphase: In metaphase the chromosomes reach their most condensed state and appear relatively smooth in outline. The chiasmata that had first appeared during diplotene have now moved toward the ends of each chromosome (terminalization), leaving only the single terminal attachment between the formerly paired arms of homologous chromosomes. These remaining chiasmata prevent the separation of homologous chromosomes which now lie at the equitorial plate of the spindle attached by their respective centromeres towards opposite poles.

In cells consisting of a diploid number of four chromosomes, or two pairs of homologues, the meiotic events leading to reduction in chromosome number can be summarized as follows :

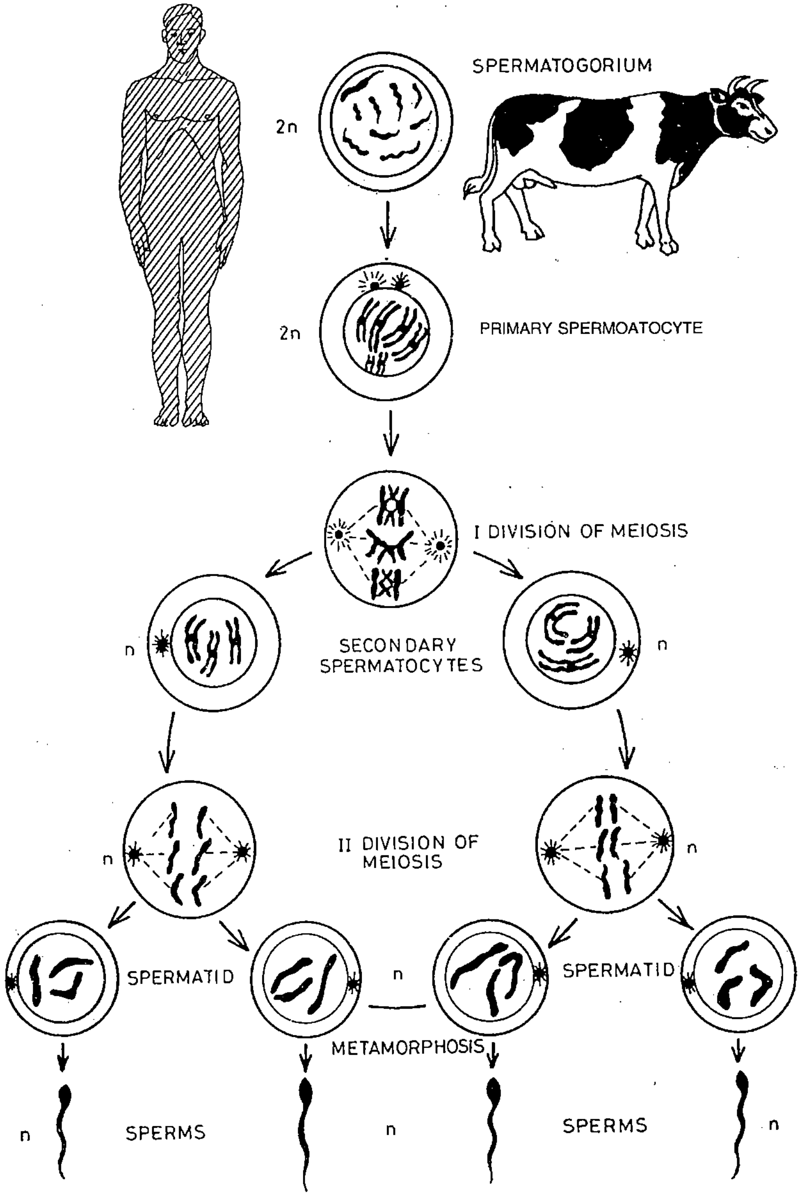
First meiotic division:

Metaphase I -	two bivalents or tetrads (four chromosomes or eight chromatids) on the metaphase spindle
Anaphase I -	two dyads (two chromosomes or four chromatids) pass to each pole

Second meiotic division:

Metaphase II -	two dyads on the spindle in each daughter cell
Anaphase II -	two monads (two chromatids) pass to each pole

Spermatogenesis



Spermatogenesis:

During the period of sexual maturity in animals, the spermatozoa are formed in the testes from reproductive cells, the spermatogonia. The final spermatagonial divisions result in cells known as primary spermatocytes, which in preparation for meiosis enter upon a period of growth.

In early prophase of the spermatocyte, the chromosomes make their appearance as fine single threads, in contrast to their appearance as double threads in ordinary mitosis. The threads soon come together side by side in pairs (synapsis). One thread of each pair is maternal in origin, the other paternal. At this stage the threads frequently show granules (chromomeres) of varying sizes and shape along their length, growing them the appearance of string of beads. Pairing takes place between identical chromosomes.

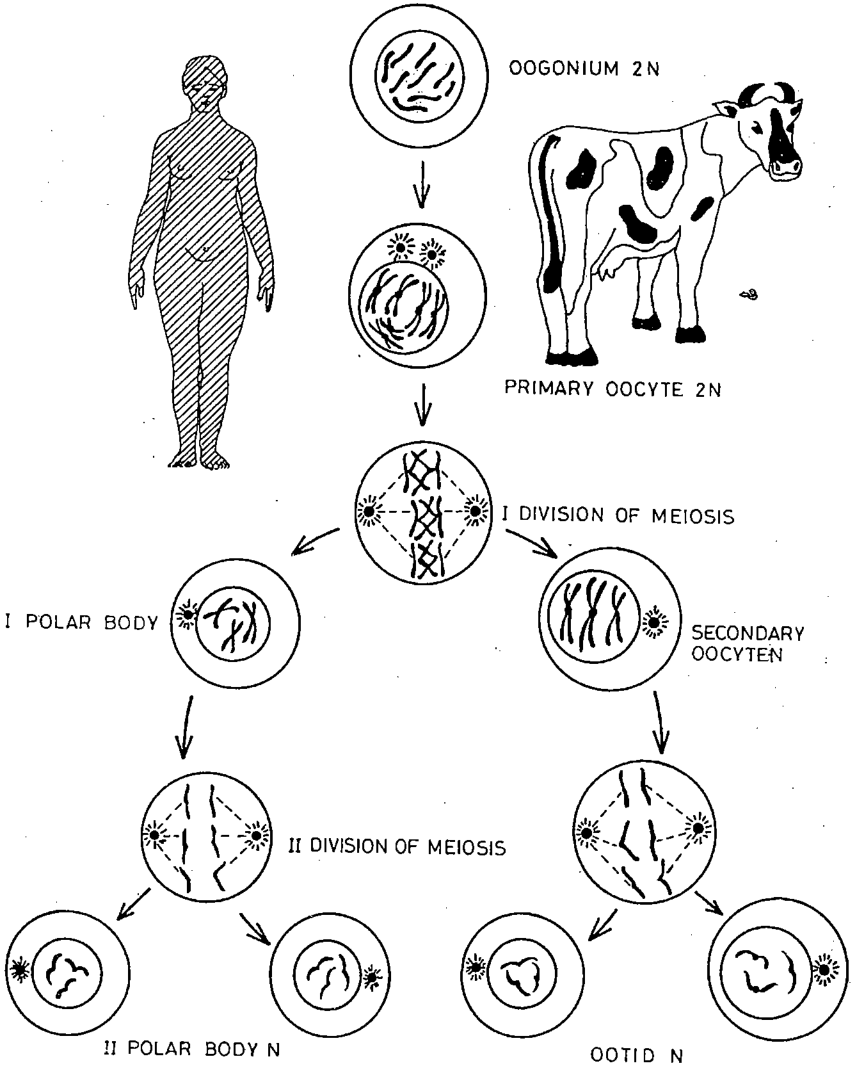
The paired chromosomes shorten and thicken, each chromosome leaving one centromere. After a time each pair seem to be made up of four half chromosomes or chromatids. The original centromeres, however, remain undivided. Groups of four chromatids are known as tetrad. The chromatids in the tetrad stage form the chiasma (Greek-meaning-crossed lines). It is at this time the genes on paternal and maternal chromatids are in a position to exchange places within one another.

The tetrads line up on the metaphase plate. The undivided centromeres, each with two chromatids, move apart and proceed to the opposite poles of the spindle. The two resulting cells, the secondary spermatocytes, undergo second division. The second division is an ordinary mitosis. The cells resulting from the second division are known as spermatids. As a consequence of the two cell divisions of meiosis, accompanied by only a single chromosomal division, each spermatid possesses just half the number of chromosomes characteristic of the species, consisting of every member of chromosome complement characteristic of the species. Spermatids undergo a process of cytoplasmic differentiation to become sperms.

Oogenesis:

A maturing egg follows a pattern similar to that of sperms. In the ovary,

Oogenesis



the immature egg, primary oocyte, grows to relatively huge size, in preparation of cell division of early embryonic development. In mammals, the first mitotic division commonly takes place before the oocyte is released from the ovary. The spindle forms near the surface and at right angles to it, so that when the oocyte divides one of the cells (polar body) receives very little of the cytoplasm, most of it being retained by the other cell, the secondary oocyte. The second division usually takes place only after the entrance of a sperm at fertilization. Again as in the first division, the partition of chromatin is equal but that of the cytoplasm is very unequal resulting the formation of second polar body. Three polar bodies remain as vestigeal eggs, being deficient in cytoplasm and eventually disintegrate. The functional egg and the polar bodies together are homologous with the four sperms.

Polytene chromosome:

In certain tissues of insects belonging to the order Diptera the cell nuclei have reached a high degree of enlargement accompanied by many extra replications of each chromosome with a single nucleus (endopolyploidy). All replicates of the same chromosome are lined up together in parallel fashion. This parallel duplication, polyteny, results in very thick chromosomes which magnify and differences in density along their length.

Lampbrush chromosomes:

The oocytes of some vertebrates with large yolk eggs expand greatly during growth period, forming correspondingly large nuclei at these stages. In some amphibia, the meiotic prophase chromosomes of such nuclei can reach 1000 microns in length, with long lateral loops giving a hairy "lambrush" appearance. Towards the end of meiotic prophase the loops begin to disappear and the chromosomes contract, so that at metaphase the bivalents are of the usual small size.

Differences between Mitosis and Meiosis :

Mitosis	**Meiosis**
An equational division separating sister chromatids.	A reduction division. The first stage is a reductional division which separates homologous chromosomes at first anaphase. Sister chromatids separate in an equational division at second anaphse.
Only one division per cycle that is, one cytoplasmic division (cytokinesis) per equational chromosomal division.	Two divisions per cycle i.e. two cytoplasmic divisions, one following the reductional chromosomal division and the other following equational division.
Chromosomes fail to synapse and no chiasmata formation.	Chromosomes synapse and form chiasmata.
Genetic exchange between homologous chromosomes does not occur.	Genetic exchange occurs between homologous chromosomes.
Two daughter cells are produced.	Four daughter cells are produced.
Genetic contents of daughter cells are identical.	Genetic contents of daughter cells are different. Centromeres may be replicas of either paternal or maternal centromeres in varying combinations.
Chromosome number of daughter cells is the same as that of mother cell.	Chromosome number of daughter cells is half of that of mother cells.
Daughter cells are capable of undergoing additional mitotic divisions.	Daughter cells are not capable of undergoing another meiotic division although they may undergo mitotic division.
Normally occurs in all somatic cells.	Occurs only in specialised germ cells.
Begins at the Zygote stage and continues throughout the life of of the organism.	Occurs only after puberty, in higher organisms. But occurs in the zygote algae and fungi.

EXERCISE

1. Write in detail on mitolic division.
2. Write briefly on spermatogenesis and oogenesis and the differences between them.
3. What are the differences between mitosis and meiosis?
4. Write shot notes on
   - a) Synapsis of chromosomes
   - b) Centromere
   - c) Diakinesis
   - d) Tetrads
   - e) Polytene chromosome
   - f) Lampbrush chromosome

Mendelian Genetics

Chapter 5

Mendelism

Basic rules of heredity were brought to light on rediscovery of Mendel's work on plant hybridization in the year 1900.

Gregor Johann Mendel was born on the 22nd of July, 1822 in Heinzendorf, a village in Silesia then a part of Austro-Hungarian Empire, now a part of the Czechoslovakia. His ancestry probably was of German and Czech. His father was a farmer and fruit grower. As a bright boy, he graduated from the high school with excellent records in 1840. But his poverty and ill health interrupted his studies and he had to seek employment to support his family. With the money he saved he completed a two year course in Philosophy.

Having been advised to take up to a less strenuous career, he entered the Augustinian monastery at Brunn in 1843 and after five years of the completion of his theological studies, was ordained as a monk on his 25th birthday.

It was customary in those days for religious heads to engage themselves in some kind of creative work in subjects like science and philosophy. Born and brought up in a farming community and having been associated with his father in the art of growing fruit plants, Mendel was much interested in doing some experiments in plants. There were in his monastery garden several varieties of ornamental plants, peas, beans, plums, pepper, maize and flax. He loved animals too. He had a pet fox and a colony of mice for breeding experiments.

Financial difficulties forced him to take up an assignment as a part-time teacher in a high school where he taught Greek and Mathematics. He studied at the University of Vienna from 1851 to 1853 taking courses in Zoology, Systematic Botany, Paleontology, Physics and Mathematics, but failed to win a certificate. However he continued his career as a teacher of Physics and Natural History in the Brunn Modern School. It was during these days, 1856-1863 he took to his experiments in crossing the edible pea plants seriously. The background of his university education gave him the ability to carry out systematic experimentation, record keeping and analysis of data. Mendel delivered his first report on his experiments before the Brunn Society for the study of Natural Sciences on February 8, 1865 in one of the classrooms where he taught. In the second meeting after a month he concluded his discussions. His paper was published in the proceedings of the society in 1866; but it lay dormant for over 34 years until they were rediscovered by the scientists in 1900.

Mendel had reputation as a popular and good teacher. He was sensitive and of an independent nature. When he was convinced of the rightness of anything he would be the most stubborn man. This is said to be a reason for his failure in the university examination. He had passed the theory but failed in the orals. He seemed to have a clash of ideas with the examiners, when he was offended and he withdrew from the examination.

He died in the year 1884. His funeral was attended by many, the rich and the poor, the lawyers, clergymen and the scientists. Little did they know at that time that herein lies the body of a man who has wrested a secret from nature to cause a revolution in the future of Genetics.

Experiments with peas:

Mendel's first interest was in producing new coloured varieties of ornamental plants by cross fertilizing the existing varieties. He was amazed at the striking regularity with which the hybrids (cross between varieties, species or races) appeared. The preliminary findings intensified his desire to do more systematic studies on hybridization. He chose the edible garden pea for this kind of experiment for three main reasons:

1. they were available in several pure breeding varieties,
2. the flowers were well protected from the influence of foreign pollen tightly closed petals and
3. the hybrids resulting from two varieties were perfectly fertile.

He purchased several pure varieties of seeds from seedsmen but selected 22 out of them from his pure breeding work. He tested their purity by the constancy of the characters in their several generations. The seven traits with contrasting pairs of characters which he has mentioned in his papers are as follows:

Trait	**Contrasting characters**	
1. Length of stem ✓	Tall (6'-7')	Short (3/4-1 1\2')
2. Shape of dried seed ✓	Round	Wrinkled
3. Colour of cotyledon ✓	Yellow	Green
4. Colour of seed coat ✓	Grey	White
5. Shape of ripe pod	Inflated	Constricted between seeds
6. Colour of ripe pods ✓	Green	Yellow
7. Position of flower ✓	Axial	Terminal

Reasons for Mendel's success:

Hybridization experiments had been done many times with plants and animals before Mendel, but not one of the earlier experiments had succeeded in discovering the law of heredity. Mendel noticed the reasons for such failures and took every precaution to avoid them.

First he realised the necessity of keeping adequate records. He kept separate records for each generation and divided the offspring in each generation into classes according to visible characteristics he had chosen and also the exact numbers of each class. He also realised that a law of cause and effect can not be deduced from a few instances.

EXERCISE

1. Give a brief outline about the life of Mendel.
2. Give an account of his experiments on garden peas and what are the reasons for his success.

Chapter 6

Monohybridization

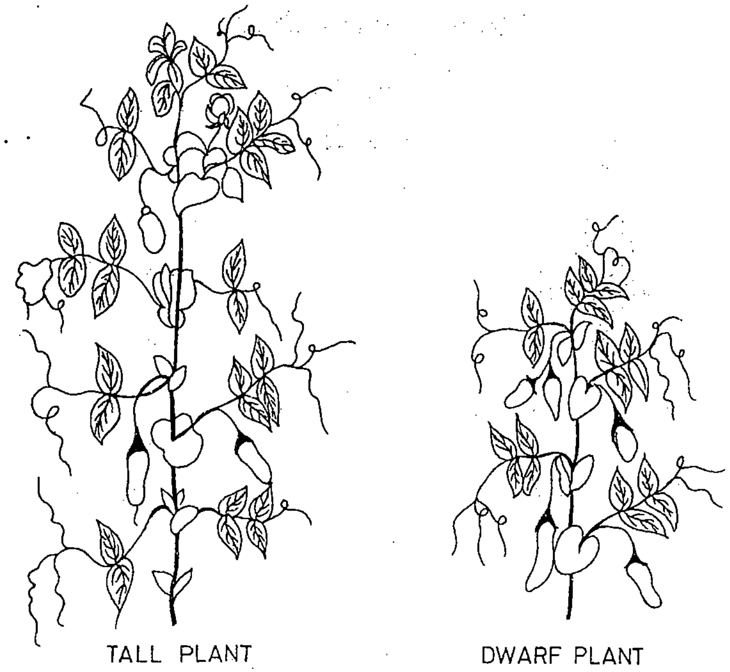
Our present knowledge of heredity rests directly on certain principles and laws discovered through the experiments and postulations of Mendel. In a little strip of garden behind the monastery building Mendel conducted his experiments on hybridization. In his experiments with plants, Mendel bred garden peas, columbines, snapdragons and many other plants, but his work on peas is ever remembered and has formed the basis of Modern Genetics.

Mendel found that when peas of the ordinary tall variety were artificially crossed with those of a dwarf variety, the resulting offsprings grew tall, like the tall parent. The original parents were termed as the "Parent Generation" (P). The tall offsprings (hybrids) were called the "First Filial Generation" (F_1), the word 'filial' from the Latin meaning 'progeny'. By self-fertilizing the F_1 tall plants, Mendel obtained 787 plants of tall variety and 277 of the dwarf variety, making approximately the proportion of 3:1. On further breeding, the F_2 dwarfs produced pure dwarf plant, while the F_2 tall ones gave rise to two kinds of offsprings. One third of them were pure tall like their pure tall grand parents, and 2/3rds of them were hybrid tall like their parents giving in turn again the proportion of three tall to one dwarf, when interbred, as summarized below:

Parents (P)	TALL (pure)	x	DWARF (pure)
F_1		TALL (hybrid)	
	TALL (hybrid)	x	TALL (hybrid)
F_2	3 TALL : 1 TALL 2 TALL (pure) (hybrid)		1 DWARF 1 DWARF (pure)

The character of height (i.e) tall and dwarf forms a pair of Mendelian character (unit character). The members of such a contrasting pair of a Mendelian character are termed allelomorphs or alleles. Symbolising 'D' for tallness and 'd' for dwarfness, 'D' and 'd' are called alleles of the unit character. Alleles that are alike, having similar pairs of genes like DD, dd, are said to be homozygotes, while an allelomorphic pair made up of unlike members as Dd, is termed as Heterozygotes or a hybrid.

Mendel's experiments with hybridization with respect to a single trait is called Monohybridization. The individuals of F_1 are called Monohybrids.



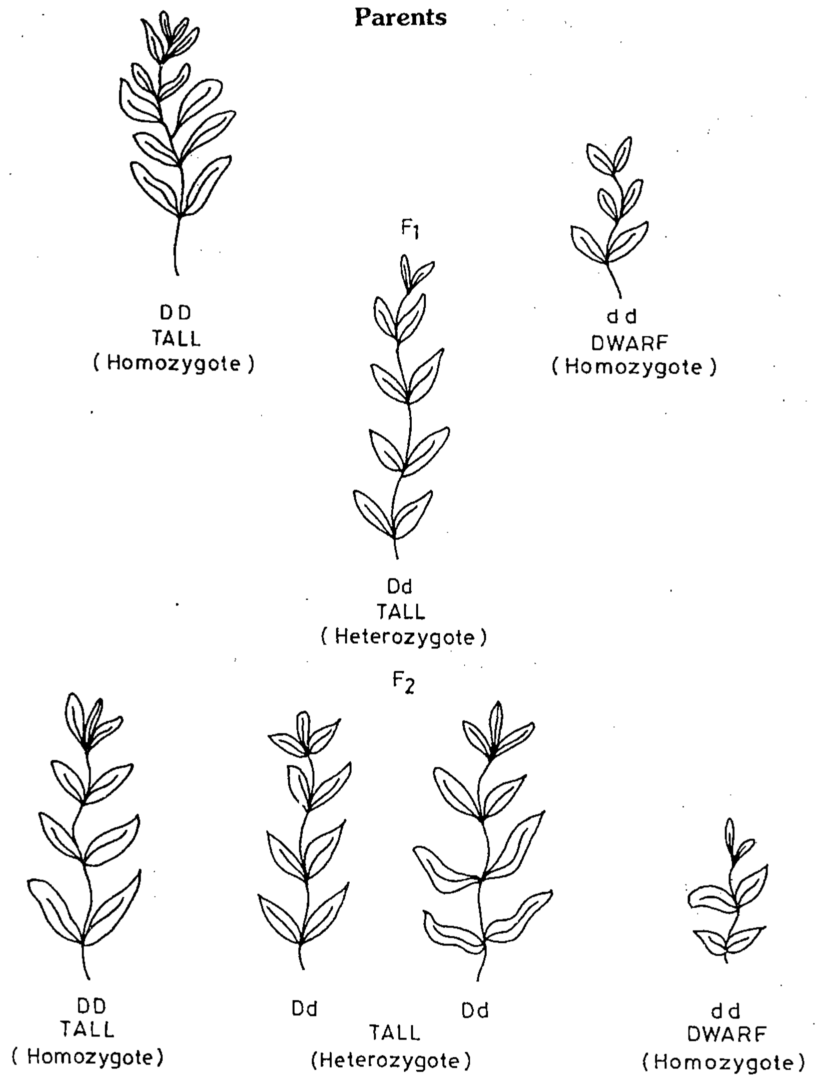
Mendel's Monohybridization—*Experiments on garden pea plants*
Tall plant - grow upto 6 ft height; Dwarf plant - grow upto $1\frac{1}{2}$ ft height
Tall character is dominant; Dwarf character is recessive

A cross between tall and dwarf varieties of plants may be summarized as follows:

Parents	TALL DD		x		DWARF dd
GAMETES	D				d
F_1		TALL Dd	x	TALL Dd	
GAMETES		D	d	D	d

Mono Hybrid Cross

[Garden Peas]



F_2 Checker Board method:

	D	d
D	DD (tall)	Dd (tall)
d	Dd (tall)	dd (dwarf)

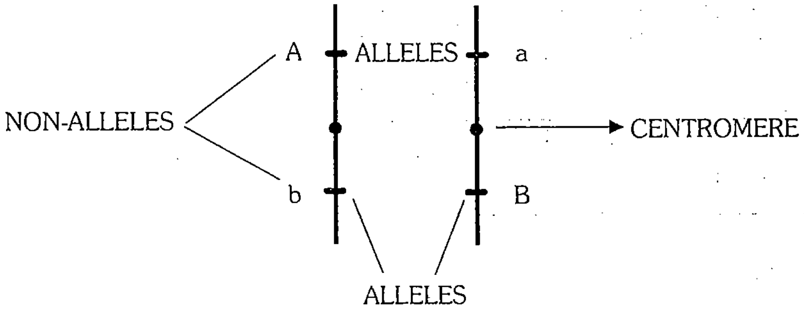
Genotypes	Phenotypes
DD -1) DD -2)	Tall
dd -1)	Dwarf

Now the question arose as to 1. What happened to the dwarf character in the F_1 generation plants? 2. How did it come back in the plants of the F_2 generation? and 3. What is the explanation for the 3:1 ratio of tall to dwarf plants in the F_2 generation?

Mendel postualated that both the parents contribute equally the factors of heredity to the offspring. The mature germ cells (gametes), the sperm and the ovum serve as the connecting link between the parents and the offspring and their union form the Zygote (union of two gametes), the origin of the new individual. Each gamete carried in it a unit factor, (now known as gene) which determines a character in the new individual.

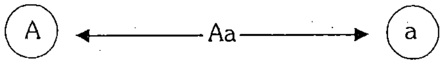
In the zygote which is formed of two gametes, these factors are in pairs, so also in all the cells of the adult individual. Cells of pure parent varieties carry similar gene pairs for the character. Symbolising 'D' for tallness, the cells in the tall variety of plants will have a pair of these genes and they are designated as `DD'. They give rise to gametes which have only one of the pairs `D'. Similarly the dwarf varieties have cells with similar gene pairs `dd' and they give rise to gametes of one kind `d'.

ALLELES AND NON-ALLELES



LAW OF SEGREGATION

HETEROZYGOTE



ALLELES A AND a SEGREGATE
TO DIFFERENT GAMETES
IN MEIOSIS

The F_1 plants in the above experiment formed by the union of the gametes 'D' from the tall parent and 'd' from the dwarf carry in their cells dissimilar gene pairs 'D' and 'd' and they are of the constitution 'Dd'. They are all tall because the factor 'D' is dominant over its other member of the pair 'd'. So the character which appeared in the hybrid, he called it the 'dominant' character. Since the factor 'd' failed to express its character namely dwarfness, in other words since it receded he called it the recessive factor and the character the recessive character.

In the cross of tall and dwarf plant, all the F_1 offspings were tall, due to the dominant factor 'D'. But still 'd' the factor for dwarfism was closely associated with it without expressing itself. The dissimilar gene pairs of F_1 (heterozygous) Dd remain in intimate union in the cells. But at the time of reproduction they segregate out in the gametes in a pure form. They give rise to two different kinds of gametes 'D' and 'd' in equal numbers with equal chances of fertilization of the same or the other kind of the gamete which explains the 3:1 ratio of the tall and dwarf plants in the F_2 generation. This is called the Law of Segregation. Different paired factors of characters derived from the two parents, although they may be intimately associated together, yet retain their individuality, separate out uncontaminated by each other and are able to form new combinations, when they unite to make a zygote.

When two contrasting characters had been brought together in the F_1 hybrid, one dominated the other in expression, in the F_2 generation, however they were sharply separated one from the other again and were distributed to different gametes.

Genetic constitution expressed and latent (DD, Dd or dd) of an organism is spoken of as the genotype while the manifested character (tall or dwarf) which is observed is known as Phenotype.

Identification of Heterozygote:

It is not always possible to distinguish a homozygote from a heterozygote and the only sure way to identify the heterozygote is by breeding it to a double recessive. This test is known as the "Test Cross" or the "Back Cross". If the individual to be tested is a homozygote, then the entire progeny of F_1 will exhibit the dominant character (tall), but if the individual to be tested is a heterozygote, then only half of the F_1 will show the dominant character and the other half, the recessive. Hence, the back cross or test cross will enable to identify the heterozygous individuals.

HOMOZYGOUS	HETEROZYGOUS
DD x dd	Dd x dd
D d	D d d
Dd	Dd dd
(all are tall)	(tall) (dwarf)
	(50%) (50%)

Mendel carried on further experiments with peas using other characters and obtained practically similar results in every case. The results are given below:

CHARACTER	Number of Dominants	Number of Recessives	Phenotypic Ratio of F_2
Form of seed	5,474 Smooth	1,850 Wrinkled	2.96:1
Colour of Cotyledons	6,022 Yellow	2,001 Green	3.01:1
Length of Vine	787 Tall	277 Dwarf	2.84:1
Colour of Flowers	705 Purple	224 White	3.15:1
Position of Flowers	681 Axial	207 Terminal	3.14:1
Form of Pods	882 Inflated	299 Restricted	2.35:1
Colour of Unripe Pods	428 Green	152 Yellow	2.82:1
Total	14,949	5,010	2.98:1 or 3:1

Heredity is a halving process as per the first Mendelian law. Mendel tested his experiments over six successive generations and found no contamination of one allele by the other in spite of close association in the same cells of the hybrid. The purity of gene is a point of fundamental importance.

EXERCISE

1. Define the following:
   - a) Mendelian factor
   - b) Monohybridization
   - c) Allele
   - d) Homozygous
   - e) Heterozygous
   - f) Dominant character
   - g) Recessive character
   - h) First filial generation
   - i) Phenotype
   - j) Genotype
2. Define and explain Mendel's law of segregation.
3. Define segregation and explain back cross test.
4. What are the offsprings expected from the following crosses of tall and dwarf garden pea plants?

   a) DDxDd b) DdxDd c) Ddxdd
5. In guinea pig rough coat (R) is dominant over smooth coat (r). A homozygous rough coated animal is crossed with a smooth one. What will be the appearance of the F_1 and of the F_2?
6. In cattle the polled (hornless) condition is due to a dominant gene P, while its recessive allele p results in horned condition.

   A polled bull is bred to three cows A,B, and C . Cow A is horned and produced a polled calf, cow B is horned and produced a horned calf.

Cow C is polled and produced a horned calf. What are the genotypes of the bull, cows A, B and C?

7. Rose comb is dominant character and single comb is recessive character in poultry. Cross a homozygous rose-combed rooster with a homozygous single combed hen and show the offspring. If the F_1, offsprings are crossed what will be the appearance of the F_2?

8. In domestic swine, there is a dominant gene which produces white belt around the body, while the recessive allele results in an uniformly coloured body. One farmer wants to produce only belted hogs, another wants only solid coloured. Which farmer could have the easier task of establishing a pure breeding stock? Tell how each could proceed?

Chapter 2

Modified Mendelian Ratio of 3:1

Mendal chose for his final experiment, pairs of contrasting characters in which essentially complete dominance prevailed over the other. Only one kind of gene action 'Dominance' or 'Recessive' was known. In other words, the gene had perfect relationship to the expression of the character.

Later, it was found that this was not always true. Dominance was not a universal law, and it was discussed that certain characters did not permit a sharp differentiation and clear separation, since the difference was of a blending nature, often difficult to define.

In Four O'clock Plant (Mirabilis Jalapa), when a pure strain of red flowered variety is crossed with a white flowered variety, the F_1 offspring (hybrid) are neither red nor white, but, have a pink flower. When these pink flowered varieties are crossed with each other, they produce red, pink and white varieties in the ratio of 1:2:1.

Parents	Red RR	x	White rr
F_1		Rr (Pink)	
F_2	RR Red 1	Rr Pink 2	rr White 1

No. of Phenotypes - 3.

No. of Genotypes - 3.

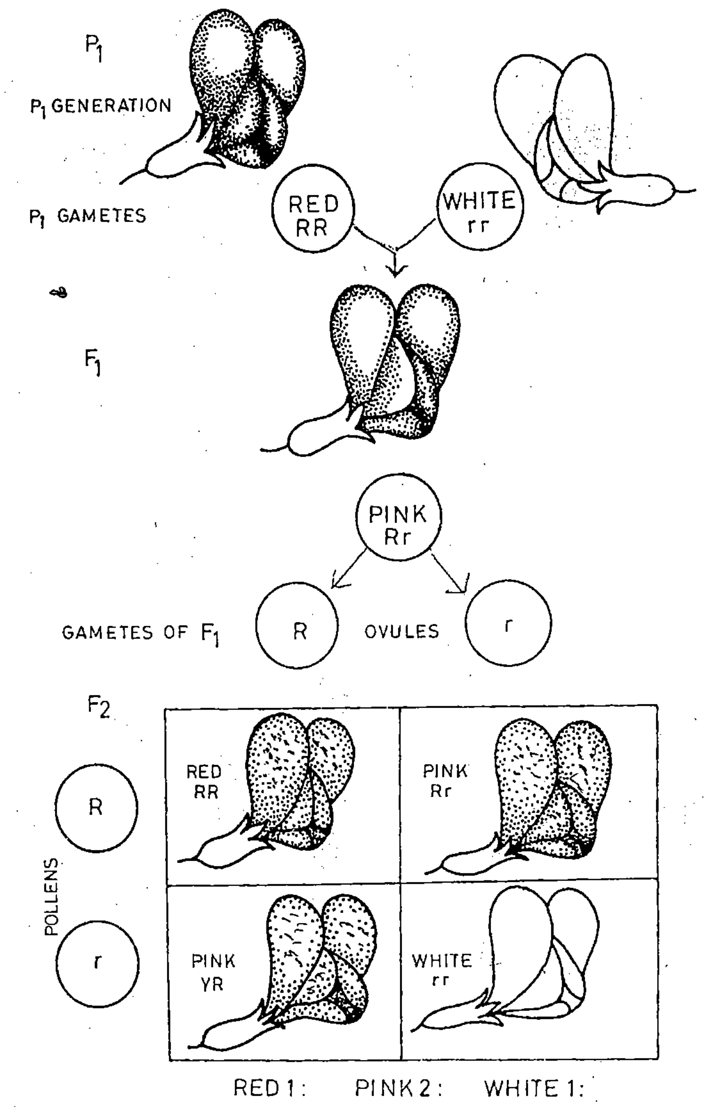
Here, the dominance is not pronounced and it is possible to distinguish the homozygotes from the heterozygotes without resorting to the back cross test. Such a type of inheritance where the heterozygous individual shows the characteristics mid way between the homozygous parents is known as Intermediate Inheritance. It is also known as partial dominance or lack of dominance or incomplete dominance according to the degree of resemblance of the heterozygote to either of the parents.

Example in cattle:

In short-horn cattle, red and white individuals exist. If a Red short-horn bull (RR) is mated with a White short-horn cow (rr) or vice versa, the offspring of the F_1 generation are Roan coloured (Rr) (a mixture of red and white hair). When two Roan (Rr) individuals are crossed, they produce red, roan and white individuals in the ratio of 1:2:1 (RR:Rr:rr). The Roan individuals have a fine mixture of red and white hair on their body, which is an expression of both the alleles, responsible for coat colour. There is no dominance of any one allele over its pair.

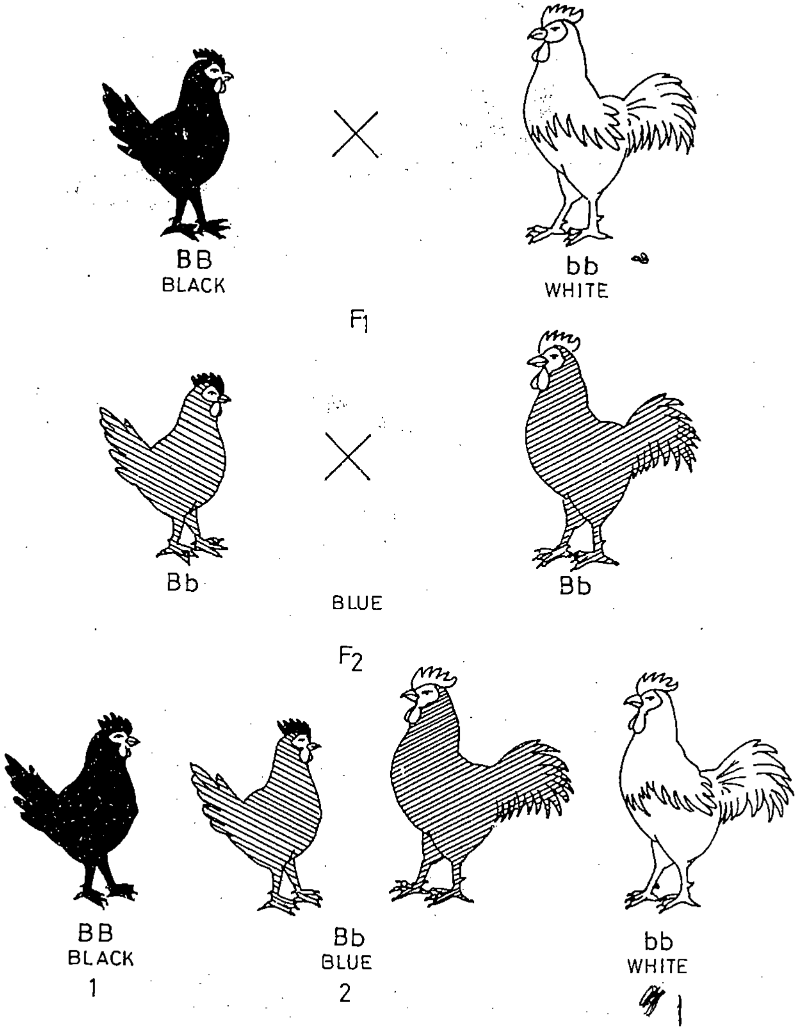
Incomplete Dominance

(Four O'Clock Plant)

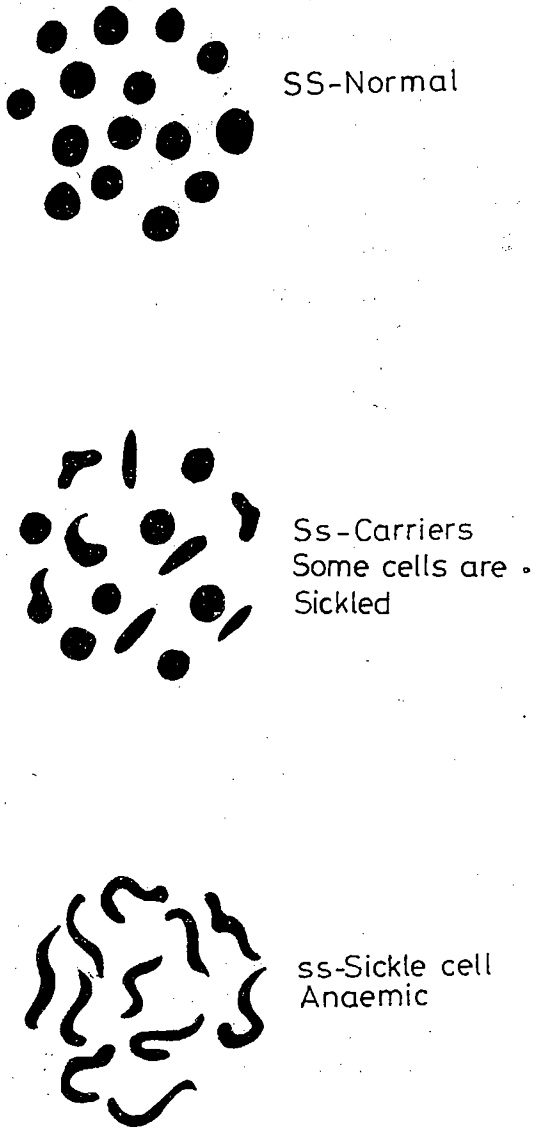


Incomplete Dominance

(Poultry)


BB
BLACK
bb
WHITE
F1
Bb
BLUE
Bb
F2
BB
BLACK
1
Bb
BLUE
2
bb
WHITE
1

Sickle cell anaemia


SS-Normal
Ss-Carriers
Some cells are
Sickled
ss-Sickle cell
Anaemic

Example in Poultry:

In poultry, when birds with splashed-white plumage (predominantly white feathers, with scattered slate blue flecks) are mated to solid blacks, the offspring are blue having slate blue feathers, edged with black as in the Blue Andalusian Breed.

Parents	Black WW	x	Splashed White ww
F_1	Blue Ww	x	Blue Ww
F_2	WW Black 1	Ww Blue 2	ww White 1

The blue chickens here correspond to the peas, symbolised 'Dd' tall (dwarf) (Dd), heterozygous for a pair of allelic genes. Neither allele is dominant. There is lack of dominance, and the heterozygotes have an intermediate blending effect, i.e. Blue.

Example in Man:

In human beings, there are numerous instances of close superficial resemblance of a heterozygote to one of the homozygous types. They could be distinguished only by special means.

A case of partial dominance is encountered in a condition known as Sickle Cell Anaemia. Sickle Cell Anaemia, a chronic disease characterised by severe chronic anaemia, retarded physical development and pain in the joints, muscles and abdomen and usually fatal. Microscopical examination of the blood smears show the red cells with gross abnormal forms like that of a sickle. The disease is due to a recessive gene 's' and the genotype of a patient with this disease will be 'ss'. Normal individuals will be 'SS'. Heterozygous individuals Ss, though appear to be healthy, when their blood was examined under the microscope, a small percentage of the red cells are seen to be sickled.

Dominance is only a relative term, and varies greatly. Complete dominance is manifested where the effect of an allele in heterozygous condition is identical with its effect in the homozygote. Where dominance is lacking or absent, the effect of one member of a pair of genes in the heterozygote is much more apparent than is the effect of its allele. This is often called Incomplete Dominance.

कृपया रेषा मारून
पुस्तक खराब करू नये.

EXERCISE

1. Define and explain on incomplete dominance with examples.
2. In short-horn cattle RR results in red coat colour, Rr results in roan and rr results in white colour. A white cow is mated to a roan bull. What are the chances of the calf to be white, roan and red?
3. A yellow guinea pig is crossed with a white one and the offspring are all cream coloured. Two cream coloured guinea pigs when crossed produce yellow, cream, and white offspring in the ratio of 1:2:1 respectively. How are these colours inherited?
4. A cattleman has a roan bull and some white cows. He wants to establish a true breeding red herd. Is it possible? If so outline a breeding programme for him to achieve his object.
5. Blue Andalusian fowls do not breed true to the blue colour of the plumage. How do you explain this? What offsprings will a blue Andalusian have if bred to birds of the following plumage colours?

   1) Black 2) Blue and 3) White

Chapter 8

Dihybrids

Monohybrids which involve a single pair of alleles are comparatively simple. But the situation becomes complicated when two organisms are crossed which differ from each other with respect to two different unit characters (Dihybrid cross).

Mendel solved this problem of dihybrid by crossing plant producing smooth yellow peas and wrinkled green peas. He found the smoothness of the form of the seed (S) was dominant over wrinkledness (s), and the yellow colour of the cotyledon (Y) was dominant over the green colour (y). If a smooth yellow (SSYY) is crossed with a wrinkled green (ssyy) all the F_1 offspring are phenotypically smooth yellow (SsYy). But they carry concealed factors for wrinkledness and greenness.

When the factors of these hybrids segregate out during the formation of germ cells they may recombine in a random manner so as to form four possible types of gametes viz. smooth yellow (SY), wrinkled green (sy) and two new combinations, smooth green (Sy) and wrinkled yellow (sY). This result is due to the fact that the two pairs of alleles are independent of each other and thus can assort into four different combinations of double gametes. This phenomenon is, known as "Random Assortment".

Since the male and female hybrids, each produce these four possible gametic combinations, the total number of zygotes that may be formed by their union will be 4 x 4 = 16, i.e., the monohybrid proportion of 3:1 which is squared, i.e., $(3:1)^2$. It does not follow, however, that the offspring in dihybrid crosses will always be 16 in number, or that they will always conform strictly to the theoretical expectation of $(3:1)^2$ i.e. 9:3:3:1. The progeny obtained will undoubtedly obey the laws of chance but greater the number of offspring, the nearer will they fall to the ratio of 9:3:3:1. The offspring of the F_2 as shown in the diagram below were grouped into four possible kinds of phenotypic and nine possible kinds of genotypic constitutions.

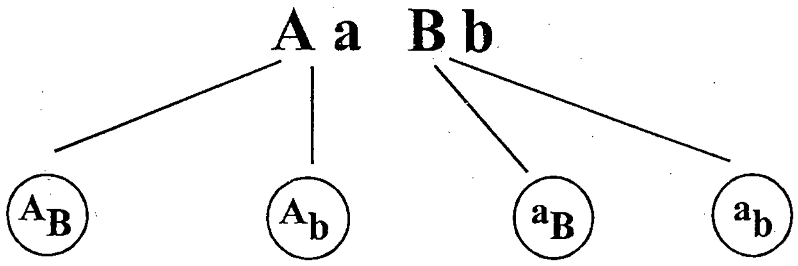
Form of Seed	Colour of Cotyledons
Smooth - S	Yellow - Y
Wrinkled- s	Green - y

Parents Smooth Yellow x Wrinkled Green
SSYY ssyy

Gametes SY sy

F_1 Smooth Yellow
SsYy

Law of Random Assortment



Two pairs of alleles have assorted independently into gametes.

Dihybrid Cross

Parents

SMOOTH YELLOW X WRINKLED GREEN
SSYY ssyy
● ⬡

$F_1 \longrightarrow$ Smooth Yellow
SsYy

♀ \ ♂	SY	Sy	sY	sy
SY	●	●	●	●
Sy	●	○	●	○
sY	●	●	⬢	⬢
sy	●	○	⬢	⬡

Smooth Yellow:	Smooth Green:	Wrinkled Yellow:	Wrinkled Green:
●	○	⬢	⬡
9 :	3 :	3 :	1

F_1 x F_1 SsYy x SsYy
Gametes Sy Sy sY sy SY Sy sY sy

F_2

	SY	Sy	sY	sy
SY	SSYY Smooth Yellow	SSYy Smooth Yellow	SsYY Smooth Yellow	SsYy Smooth Yellow
Sy	SsYy Smooth Yellow	SSyy Smooth Green	SsYy Smooth Yellow	Ssyy Smooth Green
sY	SsYY Smooth Yellow	SsYy Smooth Yellow	ssYY Wrinkled Yellow	ssYy Wrinkled Yellow
sy	SsYy Smooth Yellow	Ssyy Smooth Green	ssYy Wrinkled Yellow	ssyy Wrinkled Green

Smooth yellow 9
Smooth green 3
Wrinkled yellow 3
Wrinkled green 1

GENOTYPES PHENOTYPES

SSYY - 1)
SSYy - 2)
SsYY - 2) 9 Smooth yellow
SsYy - 4)

SSyy - 1)
Ssyy - 2) 3 Smooth Green
ssYY - 1)

ssYy - 2) 3 Wrinkled Yellow

ssyy - 1) 1 Wrinkled Green

Number of Phenotypes - 4

Number of Genotypes - 9

Considering the above two characters, smooth yellow, and wrinkled green, 3/4 show dominant smooth yellow and 1/4, recessive wrinkled green. When considered alone, they are independent of the similar segregation which takes place in other pair. This leads that 3\4 x 3\4 = 9\16 show both dominant 3\4 x 1\4 = 3\16 one of the dominant and 1\4 x 1\4 = 1\16 both recessives.

Number of Possible zygotic combinations

Pair of Genes	Number of Gametes	Possible Zygotic combinations
1	2	4
2	4	16
3	8	64
4	16	256
5	32	1024

Based on the results, Mendel rightly assumed that there is a separate factor for each character, that the factor for each character occurs in pairs in the cells and that each gamate receives one member only. All sorts of sperms have equal chance of fertilizing all kinds of eggs.

Thus Mendel obtained not only the two parental types but also the two new types. The factors do not tend to stay together in the same combinations as in the original parents but separate out and recombine in all possible combinations. In a dihybrid the characters sort out as if they have occurred in a monohybrid separately. This principle of assortment is known as Mendelian law of random assortment.

EXERCISE

1. Define law of random assortment.
2. What is the ratio of Dihybrid and discuss on Dihyhridization with example?
3. In horses black colour is dependent upon a dominant gene B, and Chestnut upon its recessive allele b. The trotting gait is due to dominant gene T, the pacing gait to its recessive allele t. If a homozygous black pacer is mated to a homozygous chestnut trotter what will be the appearance of F_1 and F_2?
4. Some dogs bark when trailing, others are silent. The barking trait is due to a dominant gene B. Erect ears (E) are dominant to drooping ears(e). What kind of pups would be expected from a cross of heterozygous erect eared barker and a droop eared silent trailer?
5. In rabbits black is due to a dominant gene B, brown to its recessive allele b, short hair is due to a dominant gene L, long hair to its recessive allele l. In a cross between a homozygous black short haired male and a homozygous brown long haired female what will be the genetic constitution and the appearance of the F_1 and F_2 generations?
6. In poultry black colour is due to a dominant gene E, red to its recessive allele e, crest head is due to a dominant gene C, plain head to its recessive allele c. A red crested male bird is mated to a black plain female. They produce many offsprings, half of which are black-crested and half of them are red crested. What are the genotypes of the parents?
7. Two cocks A and B are bred to two hens C and D. All 4 birds are feathered legged and pea combed. Cock A with both hens produce offsprings that are all feathered and pea combed. Cock B with the hen C produces both feathered and clean, but all pea combed, but with hen D produces all feathered but part pea combed and part single combed. What are the genotypes of these four birds? Feather shanks is dominant to clean shanks and pea comb is dominant to single comb.

Chapter 6

The Interaction of Genes

Sometimes from the breeding results only definite conclusions could be arrived as to the number and kinds of genes involved in a cross and their nature of inheritance. The following example will illustrate this point:

If we make a cross between a black rat and a yellow rat, we may expect in the F_1, one of three possible results. All the F_1 may be yellow, in which case we decide yellow is dominant over black or all of them may be black if that colour was dominant over yellow or yet the F_1 may be intermediate between the parents due to a blend. The actual results show that all the rats of F_1 are grey in colour. If this were to be a blend between black and yellow, in the F_2 we would expect a typical blending monohybrid ratio of one black, two grey and one yellow. But when two F_1 grey rats are crossed, four different classes of rats are produced in F_2 in the following proportions: 9 grey, 3 black, 3 yellow and 1 cream. The cream coloured rats occur in 1\16th of the F_2 individuals as a new variety, different from anything involved in this cross. This indicates that this is a two-factor or dihyrbid cross and the coat colour must be dependant not on one pair of factor but upon two pairs of factors.

Looking back at the dihybrid cross with pea, we recall that the F_1 showed the characters due to both the dominants. The 9\16 group of the F_2 also showed the characters due to both dominant genes. In this case, the F_1 were grey, representing the combined effect of a dominant gene of each pair. Similarly the cream rats occurring in 1\16 of F_2 must be due to the effect of both pairs of recessive genes. The black must be produced by the dominant gene of one pair and the homozygous recessive gene of the other. The reverse condition produces yellow.

Assuming two pairs of alleles, A and a, R and r affecting coat colour in rats and interacting as follows:

A-R	Grey
aaR-	Black
A-rr	Yellow
aarr	Cream

Homozygous Black Rat x Homozygous Yellow Rat
aaRR AArr

F1 Grey
AaRr

F_1 x F_1 AaRr x AaRr
Gametes AR Ar aR ar AR Ar aR ar

	AR	Ar	aR	ar
AR	AARR Grey	AARr Grey	AaRR Grey	AaRr Grey
Ar	AARr Grey	AArr Yellow	AaRr Grey	AArr Yellow
aR	AaRR Grey	AaRr Grey	aaRR Black	aaRr Black
ar	AaRr Grey	Aarr Yellow	aaRr Black	aarr Cream

F_2 Grey : Black : Yellow : Cream
9 3 3 1

Another interesting example of interaction of genes, denoting the influence of two genes on the same character was discovered about half a century ago by Bateson and Punnett in fowls. In poultry, there are four varieties of combs. The Wyandotte breed has a comb known as the "Rose Comb", Brahmas have "Pea Comb", while the white leghorns possess the single combs. Each of these types can be bred true. Crosses between rose combed and single combed varieties revealed that rose was dominant over single and there was a segregation of 3\4th of rose and 1\4th of single. Similar ratio was met with when a cross was made between pea combed and single combed varieties, denoting that pea combed condition is dominant over single comb.

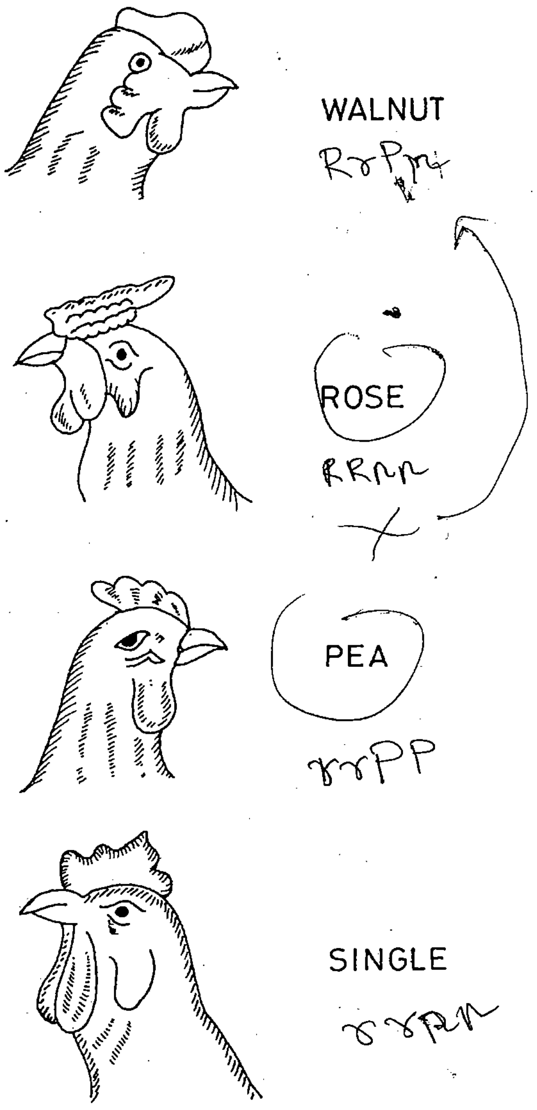
When rose combed and pea combed varieties were crossed the F_1 birds showed a new comb form known as "Walnut". The F_2 revealed four types, Walnut, Rose, Pea and Single in the ratio of 9:3:3:1. Since this is a dihybrid ratio, evidently two pairs of alleles must be involved in this cross. The single comb variety must have both of the recessive genes. This was found to be true since the F_2 singles when bred, produced only single combed progeny. The "Walnut" which is the largest class must have two dominant genes. The diagram of such a cross is shown below:

Parents Rose x Pea
RRpp rrPP

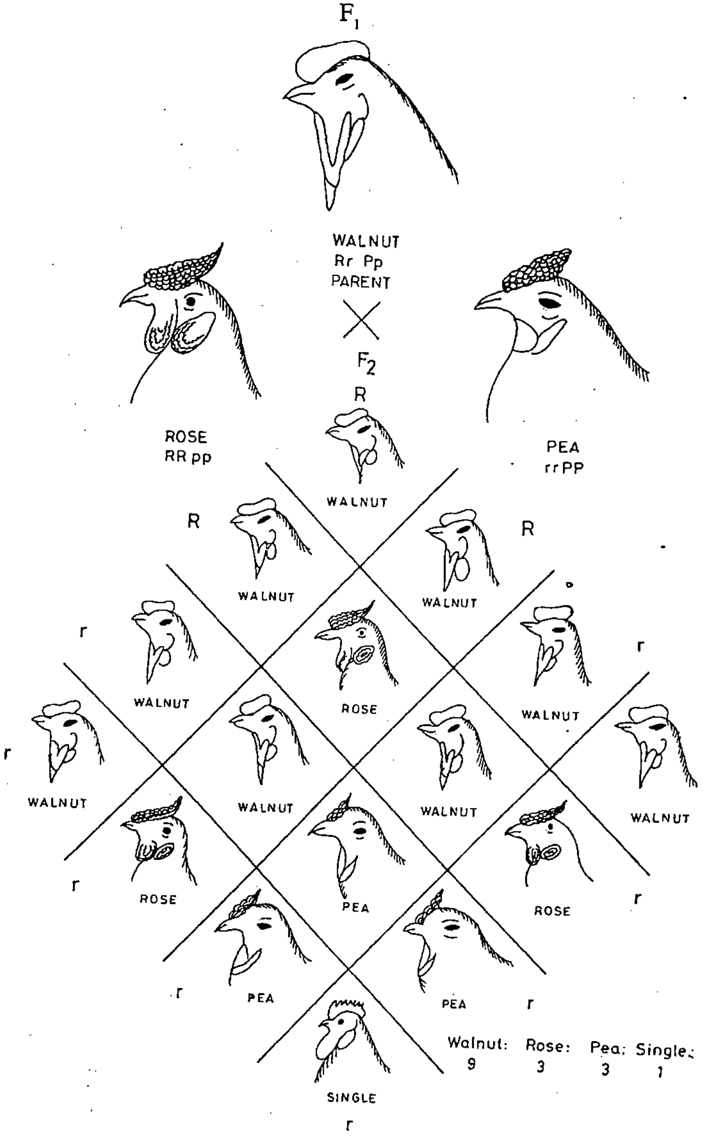
F_1 Walnut
RrPp

F1xF1

Comb Patterns in Poultry


WALNUT
RrPp
ROSE
RRpp
PEA
rrPP
SINGLE
rrpp

Interaction of Genes
Comb Pattern in Poultry


F₁
WALNUT
Rr Pp
PARENT
F₂
ROSE
RR pp
PEA
rrPP
R
R
R
r
r
r
r
r
r
r
r
WALNUT
WALNUT
WALNUT
WALNUT
ROSE
WALNUT
WALNUT
WALNUT
WALNUT
WALNUT
ROSE
PEA
ROSE
PEA
PEA
SINGLE
Walnut: 9
Rose: 3
Pea: 3
Single: 1

Gametes RP Rp rP rp RP Rp rP rp

	RP	Rp	rP	rp
RP	RRPP Walnut	RRPp Walnut	RrPP Walnut	RrPp Walnut
Rp	RRPp Walnut	RRpp Rose	RrPp Walnut	Rrpp Rose
rP	RrPP Walnut	RrPp Walnut	rrPP Pea	rrPp Pea
rp	RrPp Walnut	Rrpp Rose	rrPp Pea	rrpp Single

R-P	Walnut	9
R-pp	Rose	3
rrP-	Pea	3
rrpp	Single	1

The difference that distinguishes this and similar cases from ordinary dihybrid inheritance are that (1) that F_1 resembles neither parent and (2) in F_2 we get two phenotypes not shown in the parents.

The Walnut is the result of the interaction of the genes R and P and single comb is the result of the interaction of the recessive genes r and p. The interaction of genes is the combined action of genes to produce a phenotype which they can not produce individually. It may be an action of allelic or non-allelic pairs. The genes which interact are called as complementary genes. The R and P are non-allelic and complementary to produce Walnut comb. r and p interact to produce a single comb and they are complementary non-allelic genes.

From this we can visualise certain conclusions that could be arrived at only if actual crossing were made. The F_2 ratio of 9:3:3:1 obtained in this case only shows that certain characters are the outcome of interaction of

more than one pair of alleles. Modern genetic belief is no single gene is responsible for the production of any one character. Every character results from the interaction of all the genes, which is the basis for the modern belief of "Genic Interaction and Genetic Balance".

EXERCISE

1. Define and discuss on complementary genes with suitable examples.
2. In poultry two pairs of genes P, p and R and r interact and forms the comb as follows:

   P-R: Walnut comb

   P-rr: Pea comb

   ppR: Rose comb

   pprr: Single comb

   A pea combed hen was mated with a rose combed rooster. The offsprings produced were 2 walnut combed, 1 pea combed, 2 rose combed and 2 single combed what are the genotypes of the parents?
3. Show the expected offspring of a cross between two walnut combed chickens, both heterozygous for both genes.

Chapter 10

Modified Two Pair Ratios

The classical 9:3:3:1 dihybrid ratio may be modified in several ways. Usually four phenotypes are encountered in a dihybrid cross. Modifications of this ratio may be of two kinds i.e. modifications in which more than four phenotypes occur or less than four phenotypes.

MODIFICATIONS WITH MORE THAN FOUR PHENOTYPES

a. Lack of dominance in one pair of genes:

In short horn cattle, polled condition is due to a dominant gene 'P' while its allele 'p' results in horned condition. The coat colour is influenced by another pair of alleles 'R' and 'r' acting as follows:

RR - Red, Rr - Roan and rr - White coat.

R is not completely dominant over r. There is lack of dominance. When a homozygous polled red bull is crossed with horned white cow, the results will be as follows:

Parents Polled Red x Horned White
PPRR pprr
PR pr

F_1 Polled Roan
PpRr

Gametes PR Pr pR pr

F_2	PR	Pr	pR	pr
PR	PPRR Polled Red	PPRr Polled Roan	PpRR Polled Red	PpRr Polled Roan
Pr	PPRr Polled Roan	PPrr Polled White	PpRr Polled Roan	pprr Polled White
pR	PpRR Polled Red	PpRr Polled Roan	ppRR Horned Red	ppRr Horned Roan
pr	PpRr Polled Roan	Pprr Polled White	ppRr Horned Roan	pprr Horned White

1. Polled Red 3
2. Polled Roan 6
3. Polled White 3
4. Horned Red 1
5. Horned Roan.... 2
6. Horned White.... 1

6. Phenotypes:- 3:6:3:1:2:1

The dihybrid ratio is modified to 3:6:3:1:2:1

b. Lack of dominance in both pairs of genes:

incomplete Dominance.

For example, in snapdragon, the size of leaves are influenced by a pair of alleles B and b. There is no dominance in it, since BB-Broad, Bb-Intermediate and bb-Narrow. Flower colour is governed by a pair of alleles again lacking in dominance with the result, RR-Red, Rr-Pink, and rr-White.

A plant homozygous for both pairs of alleles, is mated with another homozygous plant.

Broad Red x Narrow White
BBRR bbrr

F_1 Intermediate Pink
BbRr
FlxF1

F_2 Gametes	BR	Br	bR	br
BR	BBRR Broad Red	BBRr Broad Pink	BbRR Int. Red	BbRr Int. Pink
Br	BBRr Broad Pink	BBrr Broad White	BbRr Int. Pink	Bbrr Int. White
bR	BbRR Int. Red	BbRr Int. Pink	bbRR Narrow Red	bbRr Narrow Pink
br	BbRr Int. Pink	Bbrr Int. White	bbRr Narrow Pink	bbrr Narrow White

BBRR	Broad Red	...	1
BBRr	Broad pink	...	2
BBrr	Broad white	...	1
BbRR	Int. Red	...	2
BbRr	Int. Pink	...	4
Bbrr	Int. White	...	2
bbRR	Narrow Red	...	1
bbRr	Narrow Pink	...	2
bbrr	Narrow White	...	1

1:2:2:4:1:2:1:2:1

9 3 3 1

9 phenotypes

9 genotypes

The dihybrid ratio is modified into 1:2:2:4:1:2:1:2:1

Each genotype gives a different phenotype. These two cases show that Mendel's F_2 ratio does not always hold good.

MODIFICATION OF DIHYBRID RATIO WHERE PHENOTYPE IS LESS THAN FOUR:

We have seen sometimes two pairs of factors or genes control the same character. In previous example of rats, where two pairs of genes affecting coat colours in rats, produced in the F_2, a ratio of 9 Grey : 3 Black : 3 Yellow : 1 Cream.

A-R-	...	Grey
A-rr	...	Yellow
aaR-	...	Black
aarr	...	Cream

It is found that in rats as well as in other animals a pair of allele 'C' and 'c' influences the pigment formation. Genotype 'CC' and 'Cc' allow the production of pigment, while 'cc' prevent the formation of pigment. Hence, 'cc' individuals are albinos. The gene 'C' is responsible for the production of an enzyme which oxidises the chromogen and produces the colour. So individuals of genotype 'cc' lack this enzyme production with the result, they are albinos; although genes for colours are present. (Albinos will have pink eyes where blood vessels can be seen through) So based on this, the coat colour in rats can be written as follows:

AA RR CC - Grey

AA rr CC - Yellow

aa RR CC - Black

aa rr CC - Cream

and the all albino rats will be aarrcc.

Recessive Epistasis

Parents

RRcc X rrcc

Black rat Albino rat

↓

F1 RrCc

Black

F2

♀ ♂	RC	Rc	rC	rc
RC	RRCC Black	RRCc Black	RrCC Black	RrCc Black
Rc	RRCc Black	RRcc Albino	RrCc Black	Rrcc Albino
rC	RrCC Black	RrCc Black	rrCC Cream	rrCc Cream
rc	RrCc Black	Rrcc Albino	rrCc Cream	rrcc Albino

Black : Cream : Albino

9 3 4

Coming back to modified dihybrid ratio, when we cross a homozygous black rat and albino rat, the cross is as follows:

Parents	RRCC	x	rrcc
	Black		Albino
F_1		Black	
		RrCc	
		$F_1 x F_1$	

F_2 Gamates	RC	Rc	rC	rc
RC	RRCC Black	RRCc Black	RrCC Black	RrCc Black
Rc	RRCc Black	RRcc Albino	RrCc Black	Rrcc Albino
rC	RrCC Black	RrCc Black	rrCC Cream	rrCc Cream
rc	RrCc Black	Rrcc Albino	rrCc Cream	rrcc Albino

Black	Cream	Albino
9	3	4

Here, we see that the dihybrid ratio of 9:3:3:1 is modified to 9:3:4. It can be seen that any rat which contains 'cc' will be albino irrespective of their possessing the colour genes 'R' or 'rr'. When a gene of one pair masks the expression of the genes of another pair, it is said to be EPISTATIC to the other pair. Because of epistasis, the number of phenotypes is reduced to three. Since, the epistatic gene 'c' is recessive to its own allele 'C', this is known as RECESSIVE EPISTASIS.

DOMINANT EPISTASIS

Another kind of modification of 9:3:3:1 ratio is seen in the following example:

Dogs: With regard to coat colour in dogs, black is dominant over brown - B is black and bb is brown. Another pair of genes 'I' and 'i' control the production of pigment with the result that animals posessing 'I' prevented from the production of colour even though specific genes for colour production is present. The difference between white and albino individuals should be known. In white animals, the eyes are pigmented unlike the albino individuals with unpigmental eyes, which look pink because of the blood showing through. For pigment production, the animals must be of the genotype 'ii' whereas 'I' genotype will be white.

Dominant Epistasis

Parents	bbii	x	BBII
	Brown dog		White dog
		↓	
F_1		BbIi	
F_2		white	

♀ ♂	BI	Bi	bI	bi
BI	BBII White	BBIi White	BbII White	BbIi White
Bi	BBIi White	BBii Black	BbIi White	Bbii Black
bI	BbII White	BbIi White	bbII White	bbIi White
bi	BbIi White	Bbii Black	bbIi White	bbii Brown

White	Black	Brown
12	3	1

When a homozygous brown dog is crossed with a homozygous white one, the resulting cross is as follows:

Parents Brown x White
bbii BBII

White
BbIi

Gametes	BI	Bi	bI	bi
BI	BBII White	BBIi White	BbII White	BbIi White
Bi	BBIi White	BBii Black	BbIi White	Bbii Black
bI	BbII White	BbIi White	bbII White	bbIi White
bi	BbIi White	Bbii Black	bbIi White	bbii Brown

White	Black	Brown
12	3	1

Here, the dominant gene 'I' is epistatic to B and b. Hence, this is known as DOMINANT EPISTASIS.

DOMINANT AND RECESSIVE EPISTASIS

Sometimes, Dominant and Recessive Epistasis occur in the same cross. White leghorns are homozygous for a dominant gene for colour 'C' but do not show colour because of the dominant gene 'I' which inhibits the pigment formation.

White silkies are also white because of homozygous recessive gene 'c' which does not produce colour and they are also homozygous for the recessive gene 'i' which allows the pigment formation. A cross between the homozygous white leghorn and white silkies is as follows:

White Leghorn x White Silkies
IICC iicc

F_1 White
IiCc
$F_1 x F_1$

Dominant and Recessive Epistasis

White Leghorn x White Silkies
IICC iicc
↓

F_1 White
IiCc

F_2

♀ ♂	IC	Ic	iC	ic
IC	IICC White	IICc White	IiCC White	IiCc White
Ic	IICc White	IIcc White	IiCc White	Iicc White
iC	IiCC White	IiCc White	iiCC Coloured	iiCc Coloured
ic	IiCc White	Iicc White	iiCc Coloured	iicc White

White	Coloured
13	3

Gametes IC Ic iC ic

F_2	IC	Ic	iC	ic
IC	IICC White	IICc White	IiCC White	IiCc White
Ic	IICc White	IIcc White	IiCc White	Iicc White
iC	IiCC White	IiCc White	iiCC Coloured	iiCc Coloured
ic	IiCc White	Iicc White	iiCc Coloured	iicc White

I - C	White	9)	
I - cc	White	3)	13
iicc	White	1)	
ii C	Colour	3	

In the F_2, 12 offsprings will inherit the dominant epistatic factor 'I' and will be white. One offspring will have 'cc', hence it will be white. Only three out of 16 offsprings will carry requisite genotype, iiCC or iiCc for colour.

In this case the dominant and recessive epistasis occur in the cross.

DUPLICATE RECESSIVE EPISTASIS

Another modification of 9:3:3:1 ratio occurs when each of the two pairs of alleles contains a recessive epistatic gene. For instance, the common yellow Daisy flowers have typical purple centres. Yellow centred varieties were discovered which when crossed with purple centred variety, gave all purple centred in F_1 and in F_2, 3 purple to 1 yellow. The F_2 yellows bred true.

Later more yellow centred flowers were discovered in another locality

and these when crossed with purple, gave the same results, 3 purple to 1 yellow. So, the yellow centred flowers of the two localities were thought to be of the same variety. But, when the two yellow centred flowers from the two localities were crossed, they produced in F_1 all purple and in F_2, 9 purple to 7 yellow. Although phenotypically, they look alike, the two yellow centred varieties were genetically different, the yellow in each case being due to a different recessive gene. To summarize the above :

Duplicate Recessive Epistasis
(Daisy Flowers)

Parents
Yellow x Yellow
PPrr ppRR
↓
F_1
Purple
PpRr

F_2

♀ ♂	PR	Pr	pR	pr
PR	PPRR Purple	PPRr Purple	PpRR Purple	PpRr Purple
Pr	PPRr Purple	PPrr Yellow	PpRr Purple	Pprr Yellow
pR	PpRR Purple	PpRr Purple	ppRR Yellow	ppRr Yellow
pr	PpRr Purple	Pprr Yellow	ppRr Yellow	pprr Yellow

Purple Yellow
9 7

Purple x Yellow Purple x Yellow
Purple Purple

3 Purple 1 Yellow 3 Purple 1 Yellow

Yellow 1 x Yellow 2

F_1 Purple

F_2 9 Purple 7 Yellow

If P and p, and R and r represent the two pairs of alleles acting in such a way that when both P and R are present the flowers have purple centres and all other combination of these factors produce yellow centres.

This is shown as follows:

Parents Yellow x Yellow
PPrr ppRR

F_1 Purple
PpRr

F_2 Gametes	PR	Pr	pR	pr
PR	PPRR Purple	PPRr Purple	PpRR Purple	PpRr Purple
Pr	PPRr Purple	PPrr Yellow	PpRr Purple	Pprr Yellow
pR	PpRR Purple	PpRr Purple	ppRR Yellow	ppRr Yellow
pr	PpRr Purple	Pprr Yellow	ppRr Yellow	pprr Yellow

Purple Yellow
9 7

Modification here is due to the recessive factor p which is epistatic to R and r, and similarly the recessive 'r' is epistatic to P and p. So this is called Duplicate Recessive Epistasis.

Another kind of gene interaction, an example of Duplicate Recessive Epistasis occurs in human beings.

In human beings deafness is hereditary. It is due to two pairs of recessive genes. We find two pairs of genes in man. A and a, B and b interact in such a way that A-B- produces normal individuals, while the recessive members aa and bb result in deafness.

When the two individuals who are heterozygous for the recessive gene pairs for deafness, the offsprings will be normal and deaf in the ratio of 9:7 as shown in figure. Even though such planned matings are not possible in human beings, analysis of pedigrees of families have shown proof of such ratio.

Parents AaBb x AabB
Normal Normal

Gametes	AB	Ab	aB	ab
AB	AABB Normal	AABb Normal	AaBB Normal	AaBb Normal
Ab	AABb Normal	AAbb Deaf	AaBb Normal	Aabb Deaf
aB	AaBB Normal	AaBb Normal	aaBB Deaf	aaBb Deaf
ab	AaBb Normal	Aabb Deaf	aaBb Deaf	aabb Deaf

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A-B- .... Normal hearing
A-bb)
aaB-) Deaf
aabb)

Such a kind of gene action where the two pairs of alleles have mutual epistatic action in the recessive condition is known as Duplicate Recessive Epistasis.

DUPLICATE DOMINANT EPISTASIS

A duplicate dominant epistasis gives a dihybrid ratio of 15:1.

Example, in poultry the shanks are normally clean and unfeathered. Certain breeds have feathers on the shanks. The Blacklanghans have feathered shanks, while the shanks of Buff Rocks are unfeathered. Assuming two pairs of factors viz. F & f, S & s, acting on shanks, the feather condition is due to the pair of alleles in their dominant state.

A cross between these breeds results in an F_1 with feathered shanks and an F_2 of 15 feathered shank birds and one with no feathers on the shanks, which occurs in a double recessive state. The ratio in this case is modified to 15:1.

Parents Feathered shanksx Unfeathered
FFSS ffss

F_1 Feathered
FfSs
F_1xF_1

F_2 Gametes	FS	Fs	fS	fs
FS	FFSS feathered	FFSs feathered	FfSS feathered	FfSs feathered
Fs	FFSs feathered	FFss feathered	FfSs feathered	Ffss feathered
fS	FfSS feathered	FfSs feathered	ffSS feathered	ffSs feathered
fs	FfSs feathered	Ffss feathered	ffSs feathered	ffss unfeathered

F-S-	) feathered - 15
F-ss	)
ffS-	)
ffss	unfeathered - 1

Only the double recessive ffss, one of every sixteen of the F_2 generation has unfeathered shanks.

Duplicate dominant epistasis is more common in plants than in animals.

DUPLICATE GENES WITH INTERACTION

The last modification of 9:3:3:1 ratio is illustrated from an example in pigs. The Duroc Jersey Pigs are usually red in colour. Sandy coloured pigs were discovered which was due to some mutation. A cross between red and sandy produced all red in F_1 and 3 red to 1 sandy in F_2. Therefore, it can be concluded that sandy proved to be recessive.

When two sandy pigs of different localities were crossed surprisingly the F_1 were all red. Obviously this means that the two sandies were not of the same genotype. Just as in the Daisies, we should expect a ratio of 9:7, in this case also.

But the F_2 results in a ratio of 9 red:6 sandy:1 white. This is a modified two factor ratio and the white animals represent the double recessive.

The two pairs of alleles R and r and S and s interact as follows:

R-S-	red
R- ss	sandy
rrS -	sandy
rrss	white

Parents Sandy x Sandy
rrSS RRss

F_1 Red
RrSs
F_1xF_1

Duplicate Genes with Interaction (Duroc Jersey Pigs)

Parents

Sandy x Sandy
rrSS RRss

↓

F_1 Red
RrSs

F_2

♀ ♂	RS	Rs	rS	rs
RS	RRSS Red	RRSs Red	RrSS Red	RrSs Red
Rs	RRSs Red	RRss Sandy	RrSs Red	Rrss Sandy
rS	RrSS Red	RrSs Red	rrSS Sandy	rrSs sandy
rs	RrSs Red	Rrss sandy	rrSs Sandy	rrss White

Red Sandy White
9 6 1

F_2 Gametes	RS	Rs	rS	rs
RS	RRSS Red	RRSs Red	RrSS Red	RrSs Red
Rs	RRSs Red	RRss Sandy	RrSs Red	Rrss Sandy
rS	RrSS Red	RrSs Red	rrSS Sandy	rrSs sandy
rs	RrSs Red	Rrss sandy	rrSs Sandy	rrss White

Red Red : Sandy : White
9 6 1

Here, the r and s genes interact and produce a cumulative effect in such a way that the rrss phenotype is different from that produced by either RRSS or rrSS. Hence, this is designated as a case of DUPLICATE GENES WITH INTERACTION. It can not be stated whether it is due to dominant or recessive epistasis and therefore is classified as Incomplete Duplicate Epistasis.

Modifications of the Dihybrid Ratio

The Results of the various types of dihybrid crosses

Genotypes	AA BB	AA Bb	Aa BB	Aa Bb	AA bb	Aa bb	aa BB	aa Bb	aa bb
A and B both intermediate	1	2	2	4	1	2	1	2	1
A intermediate B dominant	3		6		1	2	3		1
A and B both dominant (Typical dibyhrid)	9				3		3		1
aa epistatic to B or b (Recessive Epistatis)	9				3		4		
A epistatic to B or b (Dominant Epistasis)	12						3		1
aa epistatic to B or b bb epistatic to A or a (Duplicate Recessive Epistatis)	9				7				
A epistatic to B or b B epistatic to A or a (Duplicate Dominant Epistasis)	15								1
A and a interact with B and b (Duplicate Genes with interaction)	9				6				1

EXERCISE

1. What is the dihybrid ratio? Classify the modifications of dihybrid ratio.
2. Describe the modification of dihybrid ratio in which more than four phenotypes occur.
3. Define epistasis and give the ratio of F_2 generation of the following:
   a) Recessive epistasis
   b) Dominant epistasis
   c) Duplicate dominant epistasis
   d) Duplicate recessive epistasis
   e) Dominant and recessive epistasis
   f) Duplicate genes with interaction
4. In wheat red kernel colour is dependent upon the presence of two dominant genes R and B, white kernel colour upon the presence of both recessives in the homozygous state. Other combinations result in brown. Two brown varieties, rrBB and RRbb, are crossed. What is the appearance of the F_1? of the F_2?
5. In human beings two right handed parents sometimes produce a left handed child. Also two left handed parents sometimes produce a right handed child. If left handedness is heredity, could the above facts to be explained on the basis of a single pair of genes? Give reasons for your answer.
6. A yellow short haired guinea pig was crossed with a white long haired guinea pig. The F_1 were all creave short haired. The F_1 animals were repeatedly crossed among themselves. The total F_2 from many litters was as follows. 15 yellow short haired, 29 creave short haired 14 white short haired, 5 yellow long haired, 10 creave long haired, 5 white long haired. Explain these results.
7. Congenital deafness in man is due to the homozygous condition of either or both of the recessive genes d and e. Both dominant genes D

and E are necessary for normal hearing, A deaf man marries a deaf woman and all the seven children have normal hearing. What are the genotypes of the parents and children?

8. A deaf man marries a deaf woman and their 4 children are deaf. Contrast the genotypes of this family with those of previous problem.

9. In foxes nine coat colours are known as follows: Red standard silver, Alaskan Silver Double black, smoky red, cross-red, blended red, sub-standard silver, and sub-Alaskan silver. A red fox was crossed to a double black fox. The F_1 were all blended cross in colour (reddish above, black below). The F_2 showed a ratio as follows:

   1 Red: 2 smoky red: 2 Cross red: 4 Blended cross: 1 standard silver: 2 substandard silver: 1 Alaskan silver: 2 sub-Alaskan silver: 1 Double black. How many pairs of genes are concerned in the cross and is there any dominance?

10. A polled roan bull bred to a horned white cow produces a horned roan daughter. If this daughter is bred back to her father what off-spring may be expected as to horned and coat colour.

11. A homozygous polled white animal is bred to a horned red one, what will be the appearance of F_1 and of the F_2? If the offspring of a cross of the F_1 with the polled white parent? with the horned red parent?

12. In poultry white may be the result of a recessive gene O or C in a homozygous state. The presence of both the dominants is necessary for colour. The silkie breed is white and is of the genotype cc OO, the white wyandote breed was the genotype CC oo what will be the phenotypes of the F_1 and F_2 from a cross between these two breeds.

13. Rose comb is dominant to single comb in chickens. What type of off-spring would be expected from a splashed white rooster who is heterozygous rose-combed, crossed with a single combed, blue Andalusian hen?

14. In radishes the shape may be long or round or oval. The colour may be red or white or purple. A long red variety was crossed with round white variety. The F_1 were all oval purple. In the F_2 the following results were obtained. 8 long red, 15 long purple, 17 oval red, 31 oval purple, 7 long white, 15 oval white, 8 round red, 16 round purple 9 round white. What is the genetic basis for these characters?

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Chapter 11

Sex Linked Genes

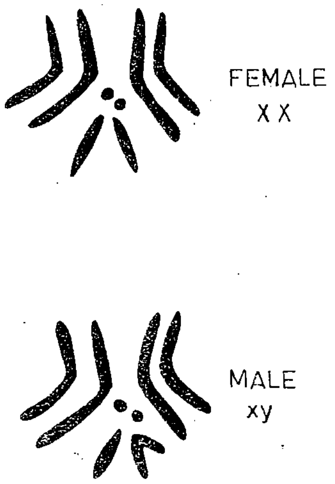
The term 'sex linkage' was used to describe the association or linkage of a hereditary trait with sex. In mammals and Drosophila, in males, sex is associated with a morphologically dissimilar (heteromorphic) pairs of chromosomes called sex chromosomes. Such a chromosome pair is labelled as x and y. Females have two morphologically identical x chromosomes. All chromosomes other than sex chromosomes are called autosomes. Genes located on the "X" chromosome (sex chromosome), are known as "Sex Linked Genes". They have a special pattern of inheritance unlike those situated in autosomes.

In 1910, T.H. Morgan, a Geneticist and a Nobel Laureate of Columbia University, working with cultures of Drosophila melanogaster, discovered the "Sex Linked Inheritance". In a culture of red eyed drosophila, he came across a white eyed male fly. This was mated with a red eyed sister. The F_1 were all red eyed. In F_2, there were three red eyed to one white eyed fly. This resembles the Mendelian monohybrid ratio with red eyed allele "W" being dominant over that of white 'w'. The strange phenomona noticed, was, in the F_2 the white eyed flies were all males. There were no white eyed females with conspicuous deviation from the normal sex ratio of 1:1. In this cross, the recessive gene apparently expressed itself only in males.

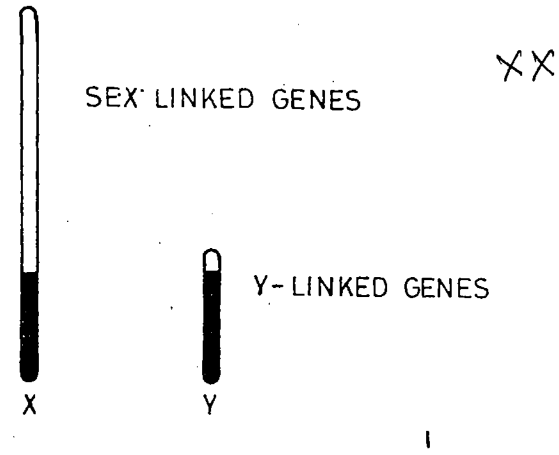
Morgan found an explanation for this on the chromosomal basis of sex determination and the sex chromosome make up of the organisms. He found in Drosophila, in the somatic cells of the male, all the chromosomes were found to occur in pairs except one without its counterpart. In females, this was found to occur in pairs with its homologous partner. This was designated as the 'X' chromosome. The other member of the unmatching pair is known as the 'Y' chromosome. In Drosophila and Man, the males are heterogametic sex i.e. XY, whereas the females are homogametic XX. Morgan hypothesised that the gene for white eyed colour follows the X (sex) chromosome pattern of inheritance. The alleles for white and red eyed were located on the X chromosome and by several types of mating this was proved to be so.

If we represent 'W' for red eyes dominant to 'w' white eyes, the white eye mutant male would necessarily carry a 'w' gene on his single X chromosome. A cross of the white eyed male with a red eyed female is shown below:

Chromosomes of Drosophila Melanogaster



Sex Linked and Y-Linked Genes



Y linked genes - also called as Holandric genes.

Hypertrichosis - long hair growth of ears is the typical example of Y linked inheritance in men.

Parents   White eyed male x Red eyed female
                 wY   WW

Gametes   w   Y   W

F_1 Red eyed female   Red eyed male
                 Ww   WY

F_1 cross   Red eyed female x Red eyed male
                 Ww   WY

Gametes   W   w   W   Y

WW   wY   Ww   wY

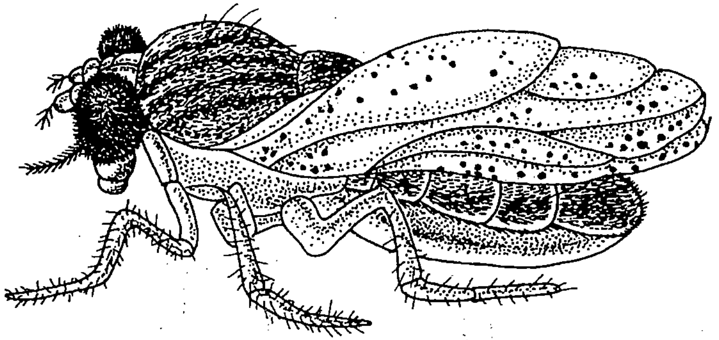
F_2	W	Y	
W	WW Red eyed female	WY Red eyed male	Red eyed female 2) Red eyed male 1) 3
			White eyed female 0)
w	Ww Red eyed female	wY White eyed male	White eyed male 1) 1

Since the male fly had only one X chromosome and an unlike Y chromosome it was considered that a single gene for white eye was capable of expression even when its allele was absent. The word hemizygous was used to describe those males that have only one member of an allelomorphic pair of genes.

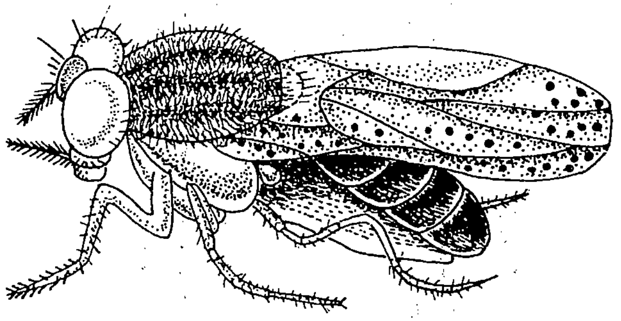
From the above diagrammatic cross, it could be seen that the 'W', 'w' alleles are carried on the X chromosomes and also in the F_2 the white eyed flies were all males. This is just what was found in the actual cross. Naturally a question arises whether it is possible to get white eyed female fly. Morgan predicted that a female of the genotype 'ww' could be produced with white eyes.

Obviously a white eyed female should be of the genetic constitution 'ww'. Hence, a white eyed female must get one white eyed gene 'w' from each parent i.e. one from the father and one from the mother. Both must carry at least one gene for this trait. So the father must be white eyed and the mother should be the carrier for this gene. Such females are available in the F_2 of the cross explained already. Crossing one of these red eyed females (heterozygous) with a white eyed male which is represented as follows:

Drosophila melanogaster



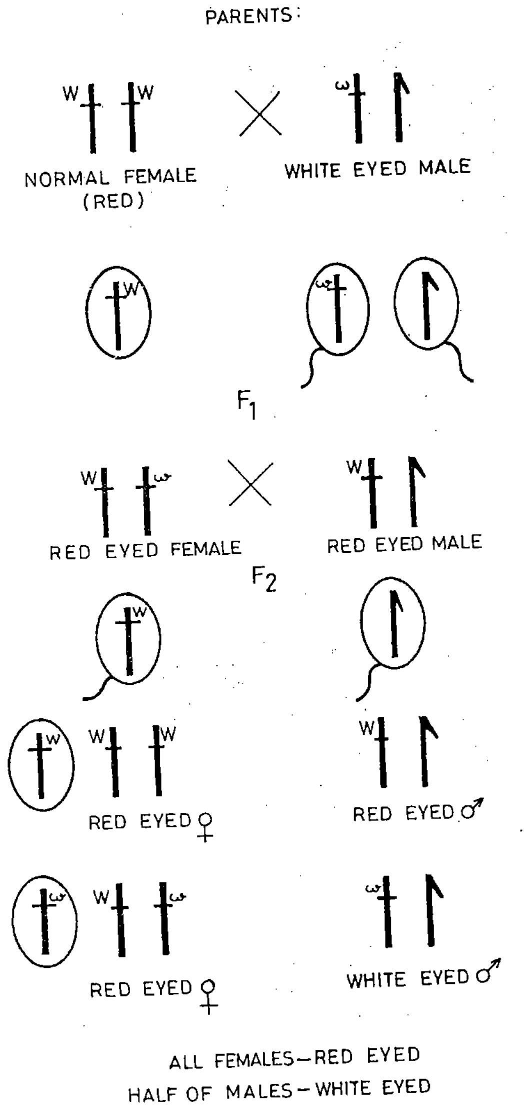
Red Eye Type



White Eye Type

Sex Linked Inheritance

[White Eye - Drosophila]


PARENTS:
W
W
NORMAL FEMALE (RED)
ω
WHITE EYED MALE
W
ω
F1
W
ω
RED EYED FEMALE
W
RED EYED MALE
F2
W
W
W
W
RED EYED ♀
W
RED EYED ♂
W
ω
W
ω
RED EYED ♀
ω
WHITE EYED ♂
ALL FEMALES—RED EYED
HALF OF MALES—WHITE EYED

Parents	White eyed male wY	Red eyed female Ww
Gametes	w Y	W w
Offspring	w	Y
W	Ww Red eyed female	WY Red eyed male
w	ww white eyed female	wY white eyed male

Half of the females were white eyed. Morgan, by actual cross of such flies proved the hypothesis that the genetic pattern of behaviour of the white eyed character clearly indicates that its gene is carried on the X chromosome.

A reciprocal cross of a white eyed female with a red eyed male is as follows:

Parents	White eyed female ww	Red eyed male WY
Gametes	w	W Y
F_1	Ww Red eyed female	wY White eyed male

F_2	w	Y
W	Ww Red eyed female	WY Red eyed male
w	ww white eyed female	wy white eyed male

Red eyed ♂ ♀ : White eyed ♀ ♂
1:1 1:1

In the F_1 generation red eyed females and white eyed males occur. This is because all the sons get only sex linked gene for white eye from the mother and the females are heterozygous red eye. In the F_2 there are equal number of red and white eyed individuals with normal sex ratio. This is a significant deviation from normal autosomal gene inheritance of dominance and recessive.

Since early times many diseases have been recorded in human beings which follow a special pattern of inheritance. According to Nasse's Law, the affected males transmit these conditions through normal daughters to about half of their grandsons. Such condition should be expected for sex linked recessive traits. The features of the sex linked recessive traits are—

1. They occur more in males than females.
2. Father and son are rarely affected.
3. It is transmitted from the affected man through the daughter to half of the grandson.
4. Does not occur in a woman unless her father has it.
5. All the sons of the women having this trait are affected.

Several sex linked abnormalities or diseases have been recorded in human beings.

1. Haemophilia
2. Colour Blindness
3. Progressive Muscular Dystrophy
4. Retinitis Pigmentosa (Progressive retinal degeneration leading to complete blindness)
5. One form of nystagmus and involuntary oscillation of the eye balls etc.

abnormal

HAEMOPHILIA (BLEEDERS DISEASE)

The primary symptoms of the disease is characterised by an abnormal tendency to bleed because of extremely slow rate of coagulation of the blood.

2-8 m

Normal blood clots in 2 to 8 minutes. In Hemophiliacs the coagulation time is greatly prolonged varying from 30 minutes to 24 hours, sometimes leading to the death of the individual.

This disease is recognised in many population, but is best known in European Royalty where there had been marriages of closely related individuals to keep up the royal status. Population studies and examination of pedigrees, indicate that it is more common in men than women. Seldom the father and son get the disease, unless the heterozygous mother passes on the gene to the son. The haemophiliac passes on the disease through the daughter to half of the grandsons. Women never get the disease, unless the father is also a haemophiliac. All sons of a haemophiliac women get the disease, since every one of them get a sex linked recessive gene from the mother.

A single dose of the recessive gene is sufficient to produce the disease in men whereas a double dose is necessary for women to produce the disease. If the incidence of the disease is 1 in 25 thousand, the chances of a woman getting the disease or getting a double dose of a sex linked recessive gene is 1/25000 x 1/25000 = 1/625000,000 or 1 in 625 million. This explains why this is more in men than in women.

COLOUR BLINDESS

This is a condition in which the individual fails to distinguish the various colours. There are several forms of colour blindness. This may be identified by means of several tests such as the colour vision test charts designed by Ishihara.

8%

In U.S.A. about 8 per cent of the men are colour blind, but among women it is only 0.5 per cent. It is more common among men than among women.

To illustrate how the mechanism of sex linked heredity is involved, we shall consider the following example:

1) If a colour blind woman marries a man with normal vision:

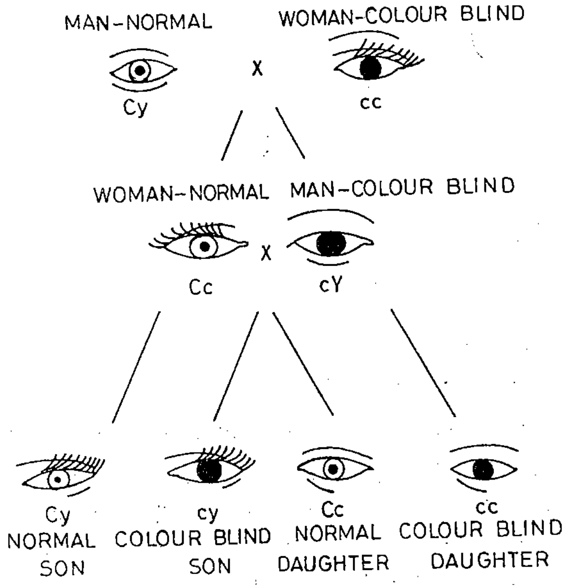
The gene for colour blindness 'c' should be present on both of the X chromosomes of a woman to be colour blind. The single X chromosome of the man should carry the normal gene 'C'. The lack of either gene 'C' or 'c' on the Y chromosome is represented as '–'.

	Mother cc	Father CY
	Colour blind	Normal
Gamtes	c	c y
	Daughter	Son
	(Normal)	(colour blind)
	Cc	cy

The reduction division produces only one kind of eggs as contrasted with two kinds of sperms. Fertilization results in the usual sex ratio of 1 male: 1 female. The daughters receive the dominant normal gene from their father while the sons are colour blind because they receive the X chromosome carrying the gene for colour blindness from their mother.

We, thus, find the sex linked inheritance peculiar in one that parents of one generation passes on the sex linked character to the opposite sex of next generation. This result is known as 'Criss Cross Inheritance.'

Sex Linked Inheritance
Colour Blindness


MAN-NORMAL
WOMAN-COLOUR BLIND
Cy
X
cc
WOMAN-NORMAL
MAN-COLOUR BLIND
X
Cc
cY
Cy
NORMAL
SON
cy
COLOUR BLIND
SON
Cc
NORMAL
DAUGHTER
cc
COLOUR BLIND
DAUGHTER

Sex Linked Inheritance
Colour Blindness

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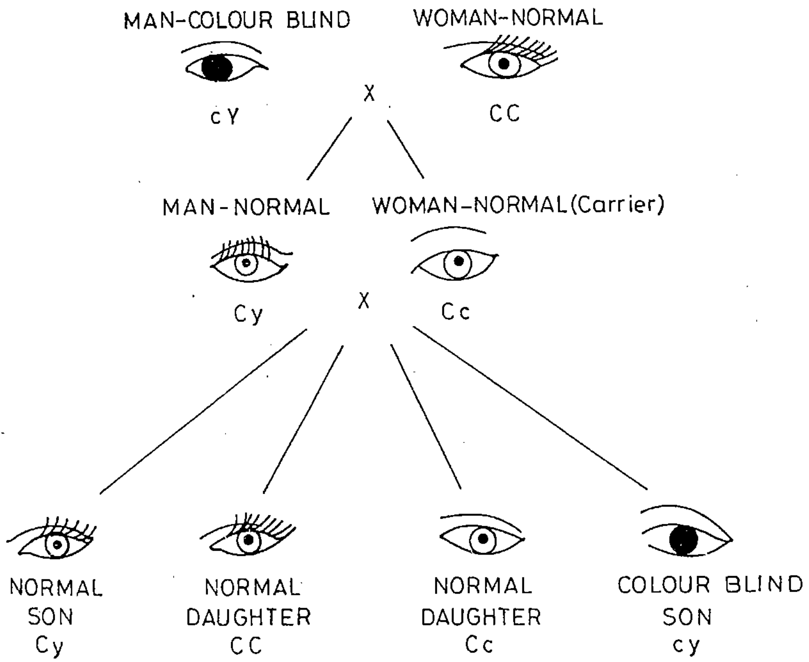
पुस्तक खराब करु नये.

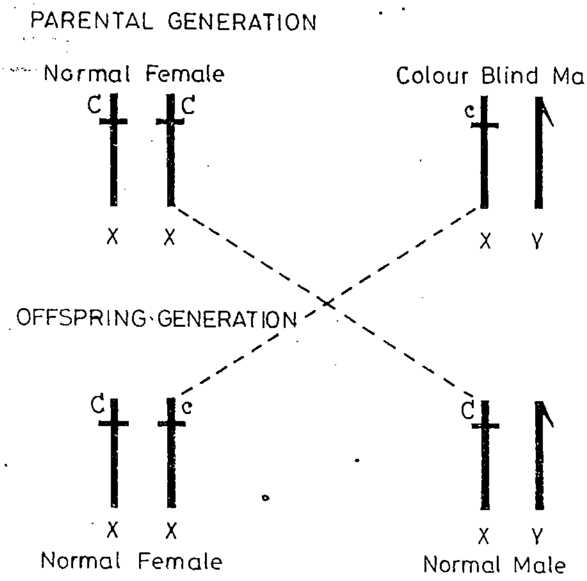

MAN-COLOUR BLIND
WOMAN-NORMAL
cY
X
CC
MAN-NORMAL
WOMAN-NORMAL(Carrier)
Cy
X
Cc
NORMAL
SON
Cy
NORMAL
DAUGHTER
CC
NORMAL
DAUGHTER
Cc
COLOUR BLIND
SON
cy

Nasse's Law : Sex linked recessive gene is transmitted from male to half of the number of grandsons through carrier daughter.

Sex Linked Inheritance

Criss-cross Inheritance

Colour Blind



Male – transmits the gene to female
Female - transmits the gene to male

Three other types of mixed matings involving colour blindness are possible. These are as follows:

2) Normal woman marries a colour blind man:

Mother (Normal)	Father (Colour Blind)
CC	cY
C	c Y
Normal Daughter	Normal Son
Cc	CY

3) Carrier woman marries a colour blind man:

Mother (normal)	Father (colourblind)
Cc	cY
C c	c Y

Normal daughter	Normal son	Colourblind daughter	Colourblind son
Cc	CY	cc	cY

4) Carrier woman marries a normal man:

Mother (normal)	Father (normal)
Cc	CY
C c	C Y

Normal daughter	Normal son	Normal daughter	Colourblind son
CC	CY	Cc	cY

Sex linked dominant gene in the domestic fowl:

It should be remembered now that sex in poultry follow the ZZ and ZW pattern, that is, the females are ZW and males are ZZ. Females are heterogametic and males are homogametics just reverse of mammals and drosophila.

In poultry the barred pattern is due to a sex linked dominant gene. This occurs in several breeds like Plymouth Rock, Dominiques, Scotch grays and Cuckoo Leghorns. Knowledge of its sex linked inheritance has found practical application in distinguishing sex of newly hatched chicken at a glance, without recourse to sexing. Identification of sex is important to poultrymen to permit early culling of unwanted males.

In barred Plymouth Rock and Cuckoo Leghorns barred condition is due to a dominant condition, represented by 'B' to its allele 'b' carrying the absence of barred condition. These factors are sex linked. Since males are homogametic i.e. of ZZ pattern, it should be assumed that the sex linked genes are paired in the males and single in the females.

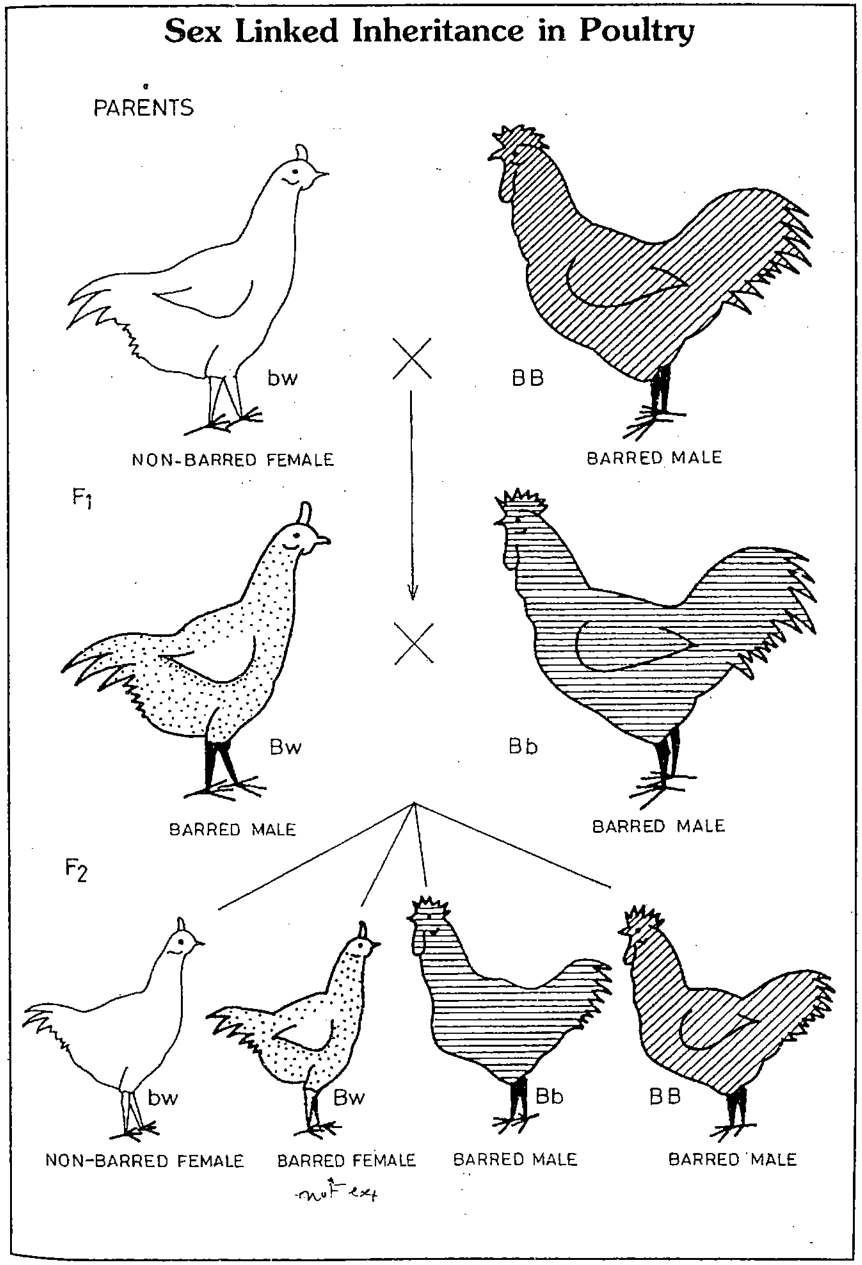
A cross between barred male and non-barred female will result as follows:

Parents		Barred male	x	Non-barred female
		BB		bW
		B		b W
F_1		Bb		BW
Gametes		B b		B W
F_2		B		b
	B	BB Barred male		Bb Barred male
	W	BW Barred female		bW Non-barred female

It is seen that F_2 gives a ratio of 3 barred to 1 non-barred but all the non-barred birds are females. This result is just opposite to that of what was seen in Drosophila. Mating of white eyed male to a red eyed female, the F_2 resulted in 3 red eyed to 1 white eyed, in which the white eyed flies are males. This is due to the reverse sex constitution in the male and female as represented by the ZZ in males and ZW in females.

A reciprocal cross of mating a barred female to a non-barred male will result as follows:

Parents		Non-barred male bb	Barred female BW
Gametes		b	B W
Offspring		b	
	B	Bb barred males	
	W	bW non-barred female	


Sex Linked Inheritance in Poultry

PARENTS
bw
NON-BARRED FEMALE
BB
BARRED MALE
F1
Bw
BARRED MALE
Bb
BARRED MALE
F2
bw
NON-BARRED FEMALE
Bw
BARRED FEMALE
Bb
BARRED MALE
BB
BARRED MALE

The male chicks received the barred factor from its mother, while the females receive no such factor from her. The male chicks as soon as hatched, were found to have a characteristic light spot on its head, later when it becomes adult gets its barred pattern. This spot on the head helps to identify the male as soon as its is hatched out. Such ~~breeds are known as~~ Auto Sexing breeds.

Lack of dominance:

Many sex linked genes appear to be completely recessive to their alleles, as in the case of white-eye gene in Drosophila and sex linked dominant genes as in the case of "barring" in poultry. Instances wherein dominance is lacking has been reported, in the case of cats. A dominant gene B represents yellow carried on the X chromosomes is allelic to b, black.

A cross between yellow female and black male cats may give the offsprings as given below:

	Yellow female	Black male
	BB	by
Gametes	B	b y
	Bb	By
	Tortoise-shelled female	Yellow male

Homozygous BB females are yellow and bb females are black, but the heterozygotes show a mixture of yellow and black, giving the so-called 'tortoise-shell' pattern. Since a male can not be heterozygous for a sex-linked pair of alleles, a tortoise shell cat is sure to be a female.

A peculiar feature of sex linked inheritance is that in F_1 the characters are inherited by the offsping in a significant manner. The male parent in 'XY' type transmits the sex linked character to the female offspring (daughter) of the F_1. From the F_1 female (daughter) this is again passed on to half of the F_2 males (grandsons). In ZW or ZO type, female parent transmits to the male of the F_1 which later pass on the character to half of the females of the F_2. We thus find inheritance becoming peculiar in that one of the parents of one generation passes on the sex linked character to the opposite sex of next generation. This type of inheritance is referred to as "CRISS-CROSS INHERITANCE".

EXERCISE

1. What is meant by the following?
    a) Sex linked genes
    b) Homogametic sex
    c) Heterogametic sex
    d) Hemizygous
2. Define the following:
    1) Criss-cross inheritance
    2) Haemophilia
    3) Nasse's law
    4) Colour blindness
    5) Autosexing
3. Drosophila white eye (w) is due to a recessive sex linked gene, the normal red eye (W) is dominant. Determine the genotypes of the parents in the three matings which produced the following offspring:

	Red eyed Female	White eyed Female	Red eyed male	White eyed male
a)	93	0	45	55
b)	100	0	0	95
c)	50	45	46	49

4. Show the expected results from a cross between red eyed male and a white eyed female Drosophila. Also show the expected results if these first generation progeny are allowed to breed among themselves.
5. A barred rooster is crossed to a non-barred hen. One half of the male chicks and one half of the female chicks are barred. The same rooster

is crossed to a barred hen. Would you expect any non-barred chicken in the offspring? Explain.

6. Show the offspring to be expected from a cross between a barred hen and a non barred rooster.

7. In the domestic fowl, silver plumage results from a dominant sex-linked gene, gold plumage from a recessive allele. Show the results of a cross between a rooster with gold plumage and a hen with silver plumage. If the first generation offspring are crossed themselves, what results would be expected in the F_2?

8. In case there is a gene for coat colour that is both intermediate and sex linked. The gene B produces yellow and its allele b produces black. The heterozygous condition (Bb) results in a peculiar combination of yellow and black and is known as tortoise-shell. Show the type of offspring expected from a cross of tortoise-shell female and a yellow male.

   ii) a black male with a yellow female.

9. A homozygous non-barred rose combed male is crossed with a barred single combed female. What kind of offspring will they expected to produce?

10. A woman with normal vision marries a man with normal vision and they have a colour blind son. Her husband dies and she marries a colourblind man. Show the types of children that might be expected from this marriage and proportion of each.

11. A normal woman whose father was colourblind marries a colourblind man. They produce a son and a daughter (a) What is the probability that the son is colourblind? (b) What is the probability that the daughter is colourblind?

Chapter 12

Sex Influenced Factors

Certain genes express the dominance depending upon the sex of the individual.

In Ayreshire cattle, the animals are spotted, either red and white or mahogany and white. Crosses between homozygous red and white and homogygous mahogany and white animals reveal an interesting and a pecular ratio:

Parents	Mahogany (Mah) and white	x	Red and white
F_1	Males	-	Mah. & white
	Females	-	Red & white
F_2	Males	-	3 Mah. & white : 1 Red & white
	Females	-	3 Red & white : 1 Mah. & white

In F_1 all the male calves were mahogany and white, while the females were red and white. In the F_2, the ratio suggests that the mahogany and white factor is dominant over red and white in the males, while in the females the reverse is the case. The reciprocal cross also gave identical results. This suggests that in this pair of alleles one seems to be dominant in males and the other in females. So in sex influenced factors, the ratios should be read twice once specifically for males and once for females. In assigning symbols to these genes, the capital letter is given for the gene dominant in males.

Let M represent the gene for mahogany and white, and m represent the gene for red and white. The genotype will show the phenotypes as follows in males and females:

Genotype	Males	Females
MM	Mah. & white	Mah. & white
Mm	Mah. & white	Red & white
mm	Red & white	Red & white

Both males and females of genotype 'MM' will be mahogany and white, since the gene 'M' can only produce this colour. Similarly, 'm' can produce only red and white in males and females. But in the heterozygote Mm, the dominance depends upon the sex. M, being dominant in males, while 'm', being dominant in females.

A diagrammatic cross between a mahagany and white male and a red and white females will be as follows:

Parents	Mahogany & White male	x	Red & white female
	MM		mm
Gametes	M		m
F_1	Mahogany & White male	x	Red & white female
	Mm		Mm
Gametes	M m		M m

F_2 males	M	m
M	MM Mah. & white	Mm Mah. & white
m	Mm Mah. & white	mm red & white

Mah. 3 & white : red & 2 white
3 1

F_2 Females	M	m
M	MM Mah. &	Mm Red & white
m	Mm Red & white	mm Red & white

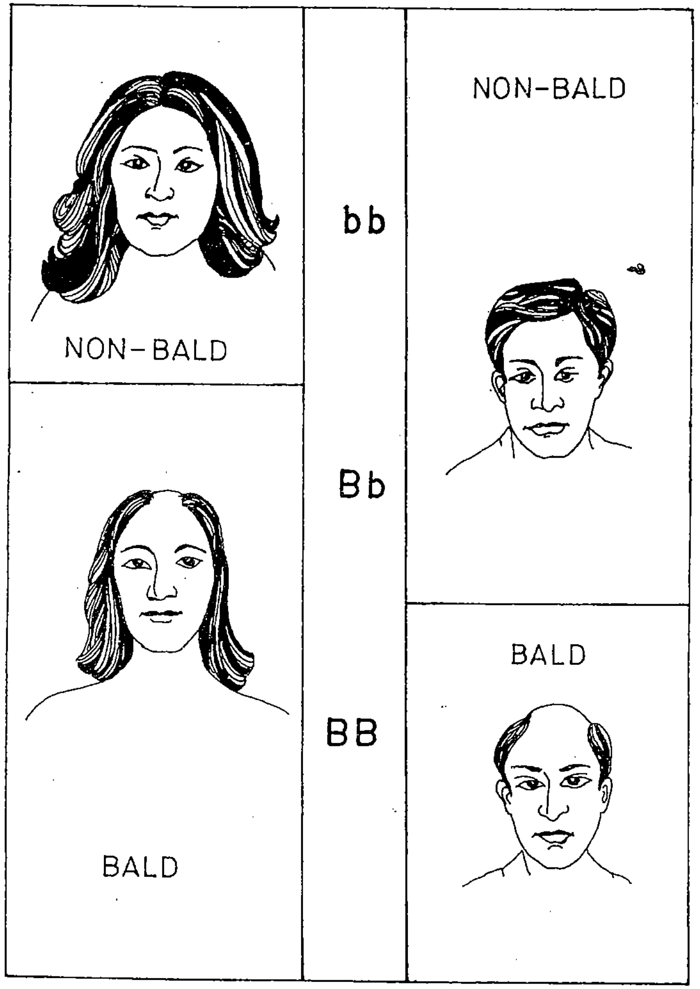
Red & White : Mah. & white
3 1

If we consider only the males, the mahogany and white act as a dominant character, since all the F_1 males are mahogany and white and the F_2 males show a ratio of three mahogany and white and one red and white progeny. Whereas in females, the red and white act as a dominant character since all the F_1 females are red and white and the F_2 females show a ratio of three red and white and one mahogany and white offspring. From the above results it is clear that in this pair of genes influencing the coat colour in Ayreshire cattle one allele seems to be dominant in males, and the other in females. In the given example, the mahogany and white gene is dominant in males and red and white gene is dominant in females.

So, in reading the ratios we should read it twice; once for the males and once for the females.

Sex Influenced Inheritance

Pattern Baldness



B - dominant in males
b - dominant in females

कृपया रेषा मारुन
पुस्तक खराब करु नये.

Example in sheep:

In sheep, the inheritance of horn follows the above pattern. In Dorset breed both sexes are horned and in the Suffolk both are hornless. A cross between these breeds gives horned males and hornless females in F_1 and F_2 among males 3 horned: 1 hornless, while the F_2 females show 3 hornless : 1 horned, indicating dominance depends upon sex.

	Males	Females
HH	horned	horned
Hh	horned	hornless
hh	hornless	hornless

Example in human:

Baldness in man may result due to diseases, seborrhoea, syphilis, thyroid disease etc. But 'pattern baldness' depends on heredity. It is more common in men than women. This leads us to think that it may be due to sex linked recessive inheritance. But the differentiation is from the fact that when a father is bald, he transmits baldness to about half of his sons, which is seldom seen in sex-linked inheritance. It is not sex linked dominant because not many women than men show the character.

If we represent 'B' as a gene for baldness, and 'b' for non-baldness, the assumption of sex influenced hypothesis is shown below:

	Males	Females
BB	Bald	Bald
Bb	Bald	Non-bald
bb	Non-bald	Non-bald

'B' is dominant in males while 'b' is dominant in females. The family records and pedigree studies indicate that, baldness is not sex linked because,

1. More common in men than women.
2. Shows up in men when neither parent show it.
3. Father and son get the trait.

Sex influenced characters are not common in animals and plants but many characters like baldness have been described in men.

EXERCISE

1. Define and discuss briefly on sex influenced inheritance with examples in cattle and sheep.
2. A Mahogany and white cow is mated to a red and white bull. Show the expected phenotypes of the sexes in the F_1 and F_2 progeny.
3. In the Dorset breed of sheep both the sexes are horned. In the suffolk breed neither sex is horned. Show the expected phenotypes of the sexes in the F_1 and F_2 when the two breeds are crossed.
4. Assume that you are a geneticist. A woman comes to you with a problem. Her mother is bald but her father is non-bald. Her elder brother is rapidly losing his hair and will soon be bald. She wants to know if she also might become bald. What would you tell her?
5. A non-bald man marries a non-bald woman. They have a son and daughter. At the age of thirty-five the son becomes bald. What are the chances that the daughter will also become bald?
6. A hornless white ram is bred to a hornless white ewe. Their offspring is a horned black male. If they produce further offspring what characters would these offspring be expected to show and in what proportions?.
7. A woman has a short index finger. Her husband has long index finger. She has five sons. What can you tell her about the length of index finger of these sons?
8. Show the offspring which would be expected in a cross between a heterozygous horned male and a homozygous horned female.

Chapter 13

Sex Limited Genes

Genes which are capable of expression in only one sex, but not in other, are known as Sex Limited Genes. In mammals and in birds, the expression of such genes depends upon the presence or absence of the sex hormones.

Beard in man is a sex limited trait. Though the genes for beard are present both in man and woman, yet shows out predominantly in man but do not express in woman. This is due to the male sex hormone that stimulates the expression of this trait.

The breast development in woman is another sex limited trait. A hormonal disturbance may cause breast development in man.

Example in cattle:

Milk production in cattle is a good illustration of a sex limited character in animals. The genes for milk are present in both the cows and bulls. The bulls also contribute their effect on the milk production of the offspring as the cows. The only means to find out the bull's ability or finding out the milk production trait in bulls is by progeny testing. When one bull produces consistently low milk producing offspring and another high milk producing offspring, we can conclude that the latter bull has genes of high milk production.

Example in Poultry:

In Poultry, plumage (feather pattern) is different in both sexes in some breeds. The feather patterns are called cock-feathered and hen-feathered. The males have a showy plumage with long feathers in the neck and tail and sickle feathers with larger comb and wattles.

In certain breeds like Seabright Bantam both male and female are henfeathered and in Campines and Hamgurghs both hen-feathered males and cockfeathered males occur.

The henfeather condition is due to a dominant gene 'H' while its allele 'h' produces cock-feather condition, which is expressed only in males. But all hens are always hen-feathered. The genotypes and phenotypes will be as follows:

	Cock	Hen
HH	Hen-feathered	Hen-feathered
Hh	Hen-feathered	Hen-feathered
hh	Cock-feathered	Hen-feathered

This indicates that in ordinary breeds of poultry where the males are cock- feathered and hens are henfeathered will be of genotype 'hh'.

Experiments reveal that the removal of testes in henfeathered males or the ovary in females results in production of cock-feathering even though 'H' gene is present. So obviously hormones control the expression of these characters.

'hh' individuals will show cock- feathering only in males and ovariectomized females.

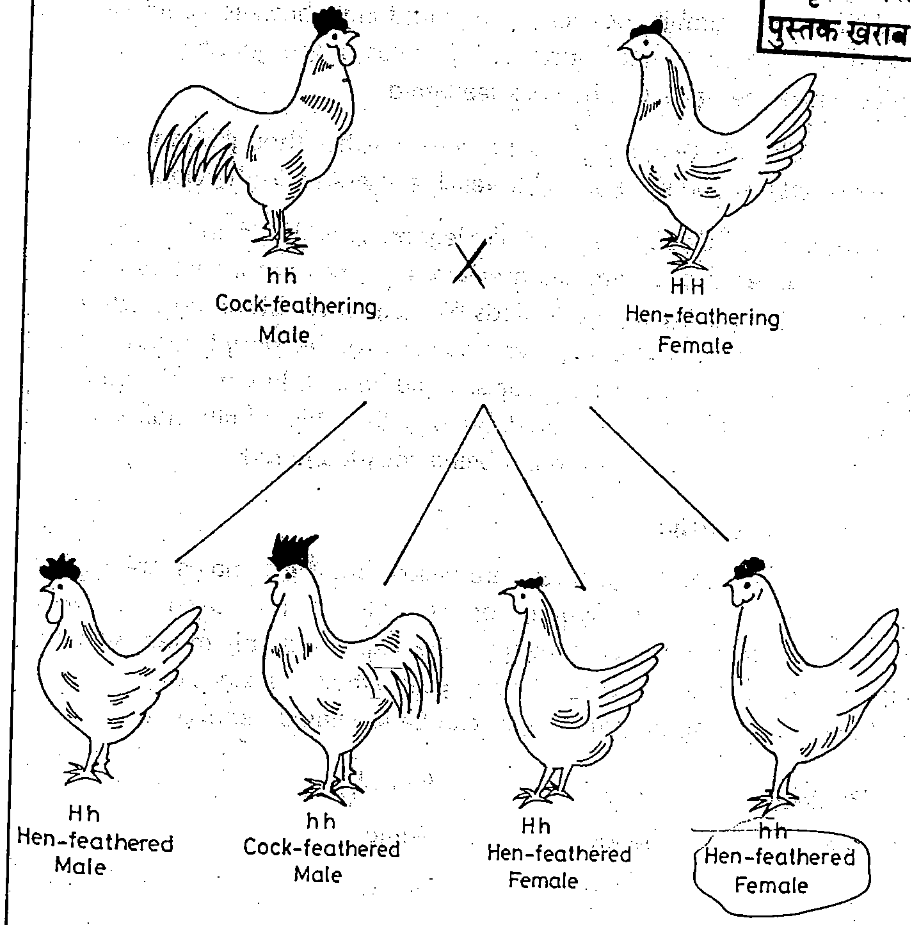
H = Hen-feathering

h = Cock-feathering

0.75

कृपया रेषा मारुन
पुस्तक खराब करु नये.

Sex Limited Inheritance



'H' inhibits cock-feathering only in the presence of the sex hormones (male or female). In the absence of sex hormone it becomes inactive (i.e.) removal of testes in a hen-feathered male or ovary in the female (HH or Hh) results in cock-feathering.

'h' does not inhibit cock-feathering but it can show its effect only in the absence of female sex hormone, i.e. males of the genotype hh, and ovariectomized females will be cock-feathered.

This indicates that certain genotypes which although homozygous expresses only in one sex and fail to reach expression in the opposite sex.

Ordinary breeds of poultry like the leghorns and R.I.R. in which males are cock-feathered and females are hen-feathered would all be of the formula-hh. Breeds having both kinds of males (hen-feathered and cock-feathered males) like the Campines and Hamburghs would contain both alleles, so that some of the individuals would be HH, Hh and hh. Here all the females would be hen-feathered, but only the HH and Hh males would show hen-feathered while hh males being cock-feathered.

Example in butterfly:

In the clover butterfly, males are yellow but the females are of two types as yellow and white. White is dominant but can be seen in females only. Letting 'W' represent a gene for white, expressed only in females and letting 'w' represent its recessive allele for yellow the occurrence of genotypes and their corresponding phenotypes can be tabulated as follows:

Genotype	Males	Females
WW	Yellow	White
Ww	Yellow	White
ww	Yellow	Yellow

The expression of character depends upon the sex.

EXERCISE

1. Define sex limited genes.
2. Write in detail on the inheritance of sex limited characters in cattle, and poultry.
3. Seabright Bantams have rose combs and hen-feathering in both sexes. Cochin Bantams have a single comb and the males are cock-feathered. If a Sebright Bantam male was crossed with a Cochin Bantam female, what would be the appearance of the F_1 and F_2.
4. A hen-feathered male bird crossed with a hen-feathered female produces 7 hen-feathered males, 2 cock-feathered males, and 8 hen-feathered females. What are the probable genotypes of the parents?

Chapter 14

Lethal Factors

Lethal factors or lethal genes produce regularly fatal effects and they modify the monohybrid ratio of 3:1. Lethal genes are those that have a drastic effect and cause the death of the young during pregnancy or at birth. Sub-lethal or semi-lethal genes cause the death of the young after birth or some time later in life.

Non-lethal or detrimental genes do not cause death but definitely reduce the viability or vigour. Lethal factors produce their lethal effect only in the homozygous condition. Lethal factors which produce a visible effect in the heterozygous condition (as in creeper fowl and Dexter cattle) are called semi-dominant lethal factors.

Lethal genes which do not produce visible effect in the heterozygous state are hard to distinguish. The only test seems to be backcross test when the homozygotes will show out.

Lethal factors were first noticed in Cambridge among yellow mice by Cuenot. When two yellow mice are crossed they produce yellow and non-yellow in the ratio of 2:1 respectively. This indicated that the yellow mice are heterozygous and yet the typical 3:1 ratio was not obtained. All the progeny with the YY genotypes would not survive.

When a yellow mouse was crossed with a non-yellow, the offspring were yellow and non-yellow in the ratio of 1:1. This resembles a back cross with yellow being dominant over non-yellow.

	Yellow	x	Non-yellow
Genotype	Yy	x	yy
Gamete	Y y		y
F_1	Yy yellow		yy Non-yellow

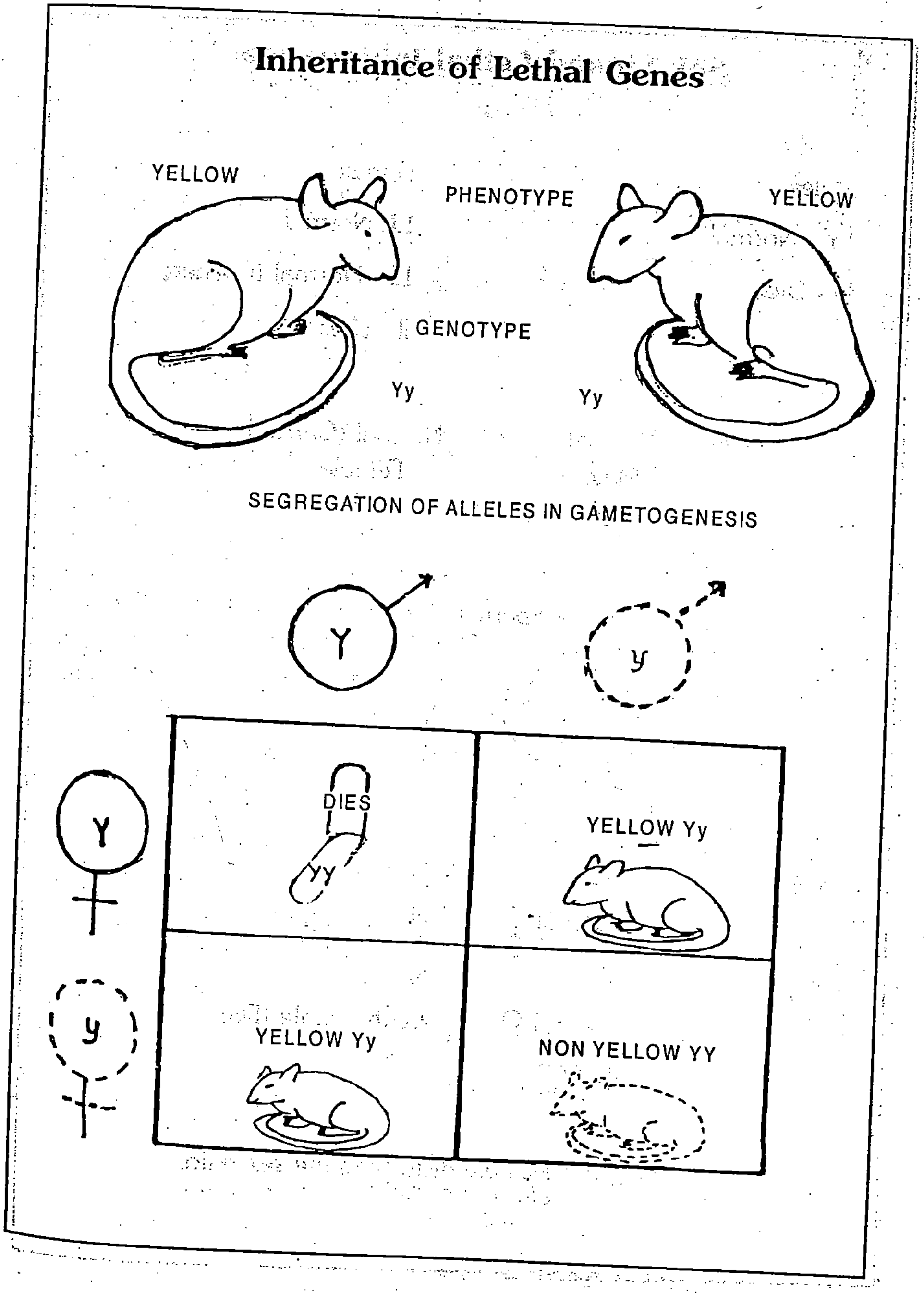
F_2 progeny

	Y	y
Y	YY dies	Yy yellow
y	Yy yellow	yy Non-yellow

When two yellow mice crossed the offsprings will be as follows:

	Yellow	x	Yellow
	Yy		Yy
Gametes	Y y		Y y

YY	Yy Yy	yy
(dies)	(yellow) Yy	(non-yellow)

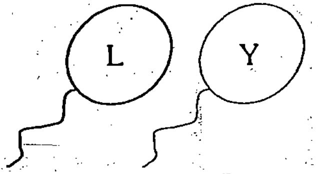

Inheritance of Lethal Genes
YELLOW
PHENOTYPE
YELLOW
GENOTYPE
Yy
Yy
SEGREGATION OF ALLELES IN GAMETOGENESIS
Y
y
Y
y
DIES
YY
YELLOW Yy
YELLOW Yy
NON YELLOW YY

Sex Linked Lethal Inheritance

Males	Females
LY - Normal	LL Normal
ly - Dies	Ll - Normal (Carrier)
	ll - Dies

Normal Male Ly x Normal (Carrier) Female Ll

Sperms



Eggs		
L	LL Normal ♀	Ly Normal ♂
l	Ll Normal ♀ (Carrier)	ly Leghal male (Die)

Half of the males die disturbing the sex ratio.

The homozygous yellow mice die i.e. 1/4 of the offspring fail to develop. The factor for yellow coat colour was therefore assumed to be a lethal factor, dominant to non-yellow with the result that hybrid yellow mice carried factors for normality and lethality. The non-yellow mice carried two normal factors, while yellow individuals carrying the lethal factors were not born at all. Analysis of the egg revealed it died on the 5th day of development. Later studies is also revealed this due to failure in the development of an enzyme. This is a case of a dominant lethal killing in homozygous state and not showing any visible effect in the heterozygous state.

Sex linked lethal gene:

When a lethal is sex linked it may readily be distinguished even though it kills early and there is no visible effect in the heterozygous state, because it will result in a marked alteration of the sex ratio. This would happen because a male of the XY type can never be heterozygous for the lethal factor and all males who inherit the lethal factor will die. The lethal gene could, therefore, be transmitted by heterozygous females. Such females while appearing perfectly normal may be discovered by the fact that they will produce only half as many male offspring as females. The females will not be killed with such a factor since in order to have the homozygous state they should receive the lethal genes from both parents, but males with sex-linked recessive lethal gene will not exist.

Let 'L' represent the factor ~~for~~ normality and 'l' for lethality and assuming that these factors are sex linked a cross may be illustrated as follows:

Males	**Females**
lY - die	ll - die
LY - normal	Ll - normal (carrier)
	LL - normal

LY x Ll

Gametes - L Y L 1

LL	L1	LY	l y
normal female	normal female (carrier)	male normal	lethal male (die)

Half of the males of the above cross will die and the significant disturbance of this sort in the sex ratio is an indication that a sex linked recessive lethal factor is operating. Haemophilia in man is an example of this condition.

In birds (poultry) the situation will be the reverse since males are homogametic (zz) and females are heterogametic (zw).

Lethal genes in Farm Animals:

Lethal genes have been recorded in cattle, horses, sheep, swine and poultry.

A few examples of lethal genes in cattle are given below:

Amputated:

A well known lethal factor known as 'amputated' is found among the Swedish Holstein Friesian cattle. This is found to occur in almost every herd. The affected calves have no appendages or if appendages are present, they are developed only to the elbows and hocks. The upper jaw is atrophied or in the form of a beak (parret jaw) and the lower jaw is almost completely absent. Cleft palate also occurs. The condition is caused by a single recessive gene. The calves are born dead. By careful analysis, it was discovered that the character amputated was inherited as a Mendelian recessive, the dominance of normality is complete. Whenever a bull and cow which are both heterozygous are mated, the amputated condition shows out.

Aa x Aa

A a A a

AA Aa Aa aa

(Normal) (Amputated)

The heterozygote did not show any visible effect. Accurate record keeping easily identified the condition and this condition arose from a bull Gallus. When two descendants of Gallus were mated, amputated condition was produced. In Gallus the mutation has occurred.

Achondroplasis in Cattle

Dexter Breed

Dexter Bull X Dexter Cow

Kk Kk

sperms

	K	k
K Eggs	KK Kerry	Kk Dexter
k	Kk Dexter	kk Bull Dog (Dies)

Kerry : Dexter : Bulldog

1 : 2 : 1

Kerry Calves - Normal

Dexter calves - Short Limbed

Bull Dog Calves - Very short legs, Cleft Palates, Short vertebral columns.

K - Normal gene

k - Lethal gene

Hypotrichesis Congenita:

This lethal condition is found in the Swedish Holsteins and in Jerseys in the United States. The hair is present only on the muzzle, eyelids, ears, pasterns and end of the tail. The body is without the initial covering of hair.

Achondroplasis-1:

In Britain the 'Dexter' show an interesting lethal condition. The Dexters are derived from another breed called 'Kerry'. The Kerry breed is a normal breed and the Dexters resemble the Kerry in most respects but is rather short limbed. When two Dexters are crossed a bull dog calf results.

Kk x Kk

Gametes K k K k

KK Kk Kk kk

(Kerry) (Dexter) (Bull Dog)

The affected calves have short vertebral columns, rounded and bulging forehead, cleft palates and very short legs. The bull dog calves are aborted after their death in the 6th to 8th month of pregnancy, following a pronounced accumulation of amniotic fluid. This is a semi-dominant lethal factor since the heterozygotes show a visible effect viz. the short limbed Dexter cattle.

Achondroplasis-2:

In the mountain cattle of Norway in a breed called 'Tele-mark' has been noticed a lethal factor called the 'Bull dog'. The affected calf has a face shortened like that of a bull dog and the limbs are short and bow legged. Affected calves are carried to full term and are born dead. This gene is inherited as a Mendelian recessive and has been traced back to a grand sire called Nicolas in whom probably the mutation of the gene took place. Any two descendents of Nicolas when bred, produced the bull dog calf.

Short-spine:

Noticed in the Oplandske of Scandinavian hill cattle. In the affected calves, the head and limbs are normal, but the spine is faulty. The centra are very small and the neural arches missing. The calves die in few hours after birth. Inherited as a Mendelian recessive.

Other abnormalities in cattle due to lethal genes are cerebral hernia, congenital spasms, curved limbs, harelip, umblical hernia, white heifer disease etc. In sheep, the defects due to lethal genes are amputated, dwarfism, muscle contracture etc.

The defects observed in swine are Atresia ani, Hair whorls, Hydrocephalus paralysis etc.

A well known semi-dominant lethal factor is known in poultry. There is a breed of chicken known as "Creeper" in which the wings and legs are very shortened giving the birds a squatty appearance. All long bones of the limbs are shortened. The tibia appears more affected than others. Some birds have their toes permanently curled.

The Genetics of this condition is peculiar.

1. Creeper x Normal

   Creeper : Normal

2. Creeper x Creeper

   Creeper : Normal

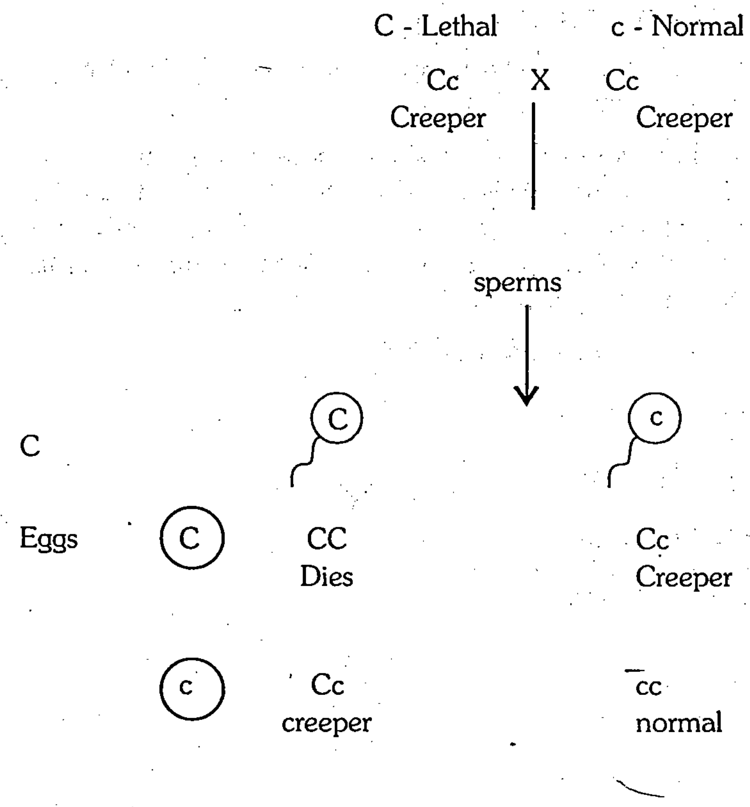
   2 1

3. Normal x Normal

   Normal

Since two normal parents never produce creeper offspring and two creeper parents frequently produce some normal offspring, it is obvious that creeper can not be due to a recessive gene. It can be due to a dominant gene since creeper is inherited directly from one generation to the next i.e. creeper bird always have at least one creeper parent. If creeper is due to a dominant gene, however, creeper birds must always be heterozygous, since creeper crossed with normal always give a 1:1 ratio. This is actually the ratio obtained by crossing heterozygous individual with a recessive individual (Test cross). Further, creeper never 'breeds true' when crossed among

Inheritance of Creeper in Poultry



Semi-Dominant Lethal

themselves since they do produce some normal offspring. Hence, creepers are always heterozygous.

When two creepers are crossed they give offspring in the ratio of 2 creepers to 1 normal, whereas it should be 3:1 under normal conditions. This is the ratio modified 1:2:1 wherein 1/4 of the embryo die in the shell around the 4th day of incubation. The results of the mating of two creepers really includes three classes of progeny, in the ratio of one non-viable embryo, two creepers and one normal. After hatching we see only the last two classes and the ratio then observed is 2 creepers : 1 normal.

Setting 'C' to represent the gene for creeper and 'c' its normal allele the cross may be diagrammed as follows:

	Creeper	x	Creeper
	Cc		Cc
	C c		C c
	C		c
C	CC dies		Cc creeper
c	Cc Creeper		cc normal

All the birds having the genotype CC would die and the resultant phenotypes of the progeny would be creeper and normal birds in the ratio of 2:1.

Lethal gene in human beings:

In human beings, several genes are suspected to be intermediate lethal. One of these in heterozygous state produce persons with short fingers, a condition known as Brachyphalangy. These persons will have only two joints instead of three joints in their fingers. X-ray studies indicate that this condition is due to the fact that the middle bone (phalanx) of the finger is greatly shortened and often fused with one of the other bones of the finger. In one such marriage of two persons with this same gene produced one child without any finger or toes and was unable to survive. Two other children showed the characteristic short fingers and one was normal. This is the exact 1:2:1 ratio which would be expected from parents heterozygous for an intermediate lethal.

Genes lethal under certain conditions:

Certain genes become lethal under certain environments. The good example is Erythroblastosis foetalis. The gene for Rh factor is not lethal. But under certain conditions it is lethal. When an Rh negative woman bears an Rh positive child she may become sensitizied to the Rh factor and generate antibodies in her system which react with Rh antigen. We generally think of heredity as a one-way process, always from parent to child, but this is an unusual case in whcih the inheritance of the child influences the nature of the parent. When such a sensitized woman bears another Rh positive child, this same reaction will occur in the body of the child causing abnormalities. Such babies are born dead or die shortly after the birth. Thus in the uterus of sensitized woman the dominant gene for the Rh factor may be a lethal.

The gene for albinism is always lethal in plants but in human beings it is not lethal under protected environment. If the albinos are exposed to intense sunlight, their skin would become so badly burned that death would result.

Frizzled feathers in chicken is another good example. This gene causes the feathers to break off easily and results in birds which are almost naked. If such birds are kept in warm environment they manage to survive but if they are exposed to cold weather, without the insulation of normal covering of feathers death occurs. Hence the gene is lethal under one environmental condition and not lethal under other condition.

Elimination of lethal genes:

Elimination of lethal genes from the population could be carried out by identifying the carriers (heterozygotes) and preventing them from further breeding. Intermediate lethal genes are much easier to detect, because all individuals carrying intermediate lethals will exhibit some phenotypic expression of the gene. As a result it is much easier to eliminate them. One must merely prevent reproduction of the easily recognizable heterozygous individuals.

EXERCISE

1. Write in detail on the inheritance of lethal genes with suitable examples.
2. Define semi-dominant lethal and discuss its inheritance with examples.
3. Write short notes on a) Sex linked lethals, b) Lethal effect under certain special occasions, c) Dexter breed of cattle.
4. What are the lethal conditions in domestic animals?. How will you eliminate them?
5. To obtain Dexter cattle breeders sometimes cross the Dexters. This cross gives one half Dexter offspring but one fourth of the calves are bull dog and die. Explain how Dexters would be obtained without any loss of the calves.
6. A Dexter bull homozygous for polled is mated to a horned kerry cow. What kinds of calves would they produce and in what proportion?
7. In poultry a gene C produces creeper condition in the heterozygous state but is lethal in the homozygous state. What kinds of offspring and in what proportions would result from the following matings:

   a) Cc x Cc

   b) Cc x Cc
8. A creeper male heterozygous for barring is mated to a creeper nonbarred female. What offspring would be expected?
9. In fowls a factor 'k' is recessive and sex linked. All zygotes pure for 'k' die before hatching. A male heterozygous for this factor is crossed with normal female and produce 120 chicks. How many of these would you expect to be males and how many females? Reason your answer.
10. A factor 'l' in Drosophila is recessive, lethal and sex linked. If female 'Ll' is crossed with a normal male, What should be the sex ratio of the progeny?

Aa Bb

AB Ab aB ab

AB Ab aB ab

The standard F_2 ratio, resulting from mating together the F_1 progeny of a cross such as AABB x aabb (AAbb x aaBB) is 9:3:3:1 i.e.

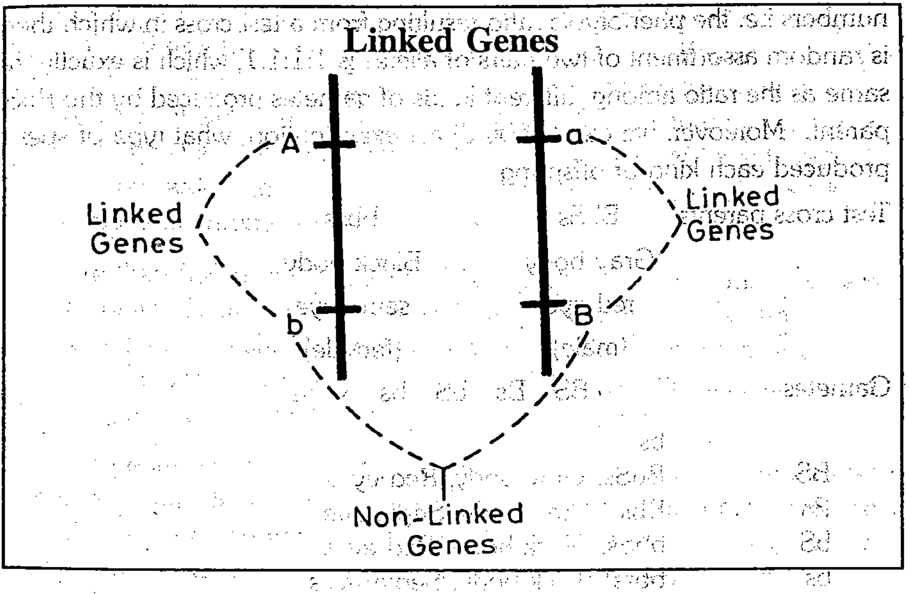
9	A-	B-
3	A-	bb
3	aa	B-
1	aa	bb

This 9:3:3:1 ratio results from the fact that the A, a alleles assort independently of the B, b alleles and that the independent assortment means that the Aa Bb individuals of the F_1 generation produce gametes in the ratio of 1AB : 1Ab : 1aB : 1ab. Obviously then, if two pairs of alleles do not assort at random then we should expect to find a disturbance of the 1:1:1:1 ratio among the gametes of individuals heterozygous for both of them (AaBb) and a consequent disturbance in the F_2 ratio.

A failure of random assortment results in the disturbance of the standard dihybrid F_2 ratio of 9:3:3:1. This will happen if the two different pairs of alleles are carried on the same chromosome pair. It is much easier to see how the two pairs of alleles are distributed among the gametes by making use of a test cross. Since a test cross is a very useful tool in genetics, let us illustrate it with a concrete example.

To start with, let us consider a case involving two pairs of genes which are in different chromosome pairs. In Drosophila numerous genes affect the colour of the body. Wild type flies have gray bodies, but there is a mutant form in which the body-colour is black. Crosses between gray and black give all gray in the F_1 and a ratio of 3 grays to 1 black in the F_2. Therefore, the genes for gray and black are allelic, with the gene for gray being dominant.

Another mutation in Drosophila changes the red eyes to sepia eyes. Again crosses between red and sepia give all red in the F_1 and a ratio of 3 red to 1 sepia in the F_2. These genes are also allelic, red being dominant over sepia.



A cross involving both black body and sepia eyes would thus be a two pair cross. Let us cross a fly which is normal except for having a black body with one which is normal except for having sepia eyes. Letting B represent the gene for gray body and b, its allele for black body and letting S represent the gene for red eyes and s its allele for sepia eyes the cross would be diagrammed as below:

Black body red eyes x Gray body sepia eyes

bbSS BBss

Gametes bS Bs

Gray body red eyes

BbSs

If the two F_1 individuals are mated the 'standard' ratio of 9:3:3:1 would be obtained. Instead, however, we shall make a test cross. A test cross is a cross in which an individual heterozygous for any number of genes (here two pairs) is mated to one which is homozygous for each recessive allele. In this case, we shall mate a F_1 male BbSs to a female double recessive, bbss. The progeny of the test cross are of four types which occur in equal numbers i.e. the phenotypic ratio resulting from a test cross in which there is random assortment of two pairs of alleles is 1:1:1:1, which is exactly the same as the ratio among different kinds of gametes produced by the BbSs parent. Moreover, we can tell by direct examination, what type of sperm produced each kind of offspring.

Test cross parents BbSs x bbss
Gray body Black body
red eye sepia eye
(male) (female)
Gametes BS Bs bS bs x bs

	bs
BS	BbSs, Gray body, Red eyes
Bs	Bbss, Gray body, Sepia eyes
bS	bbSs, Black body, Red eyes
bs	bbss, Black body, Sepia eyes

BS sperm uniting with bs egg produces a gray body red eyed fly. Bs sperm produces a gray body sepia eyed fly. bS sperm produces a black body red eyed fly and bs sperm produces a black body sepia eyed fly. Similarly we can tell directly what kind of egg from a BbSs female has produced each type of offspring from test cross in which BbSs is a female and bbss a male.

Test Cross Parents BbSs x bbss
Gray body Red Black body sepia eyes
eyes female male
Gametes BS Bs bS bs x bs
Offspring:

	bs	
BS	BbSs	Gray body, Red eyes
Bs	Bbss	Gray body, Sepia eyes
bS	bbSs	Black body, Red eyes
bs	bbss	Black body, Sepia eyes

If genes are carried on chromosomes, random assortment can happen only if the different pairs of genes are carried on different pairs of chromosomes. But if two pairs of genes are carried on the same pair of chromosomes, it would seem that they should stick together i.e. wherever each chromosome of the pair went both genes in it would have to go. To show random assortment, then two pairs of genes would have to be on different pairs of chromosomes.

More than 500 pairs of genes in Drosophila are known and there are only four pairs of chromosomes. There must, therefore be assortment can not always occur; we should find some exception to it. In these cases, the breeding result indicate, that certain genes of different allelic pairs tend to stick together in the way in which they entered the cross.

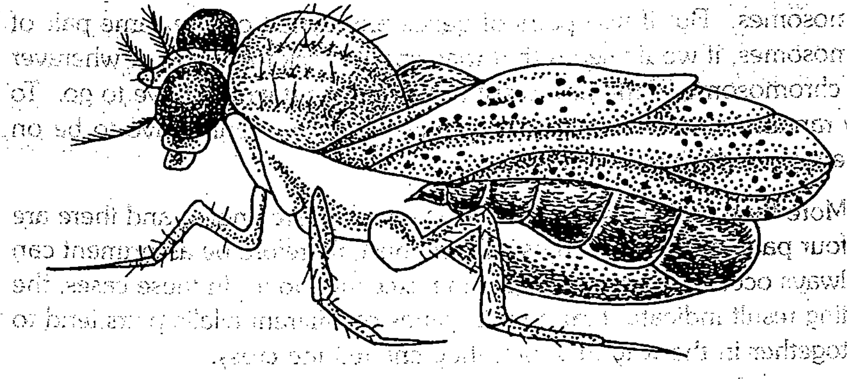
Such genes are called 'Linked genes'. Linked genes are all on one chromosome and they tend to move *en bloc* during meiosis when the chromosomes are segregated into two haploid groups. Linkage is the presence on the same chromosome of two genes affecting two traits.

Let us examine a cross in which the effects of linkage can be seen.

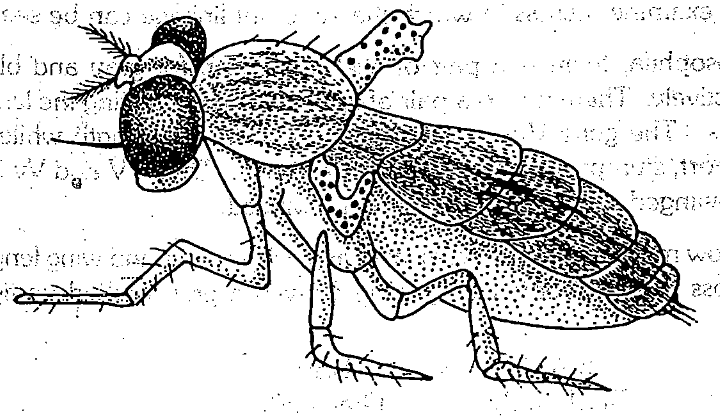
In Drosophila, there is a pair of alleles B and b for gray and black body respectively. There is also a pair of alleles V and v affecting the length of the wings. The gene V is necessary for normal wing length while vv produces short, stumpy wings known as "Vestigial". Thus VV and Vv flies are normal-winged and vv flies are vestigial winged.

If we now make a two pair cross involving body colour and wing length, we may cross a black bodied long wing fly with a gray bodied vestigial wing fly.

Parents bbVV x BBvv
Black body Gray body
long wing vestigial wing
male female
Gametes bV Bv
F_1 BbVv
Gray body
long wing



Drosophila Melanogaster - Wild Type
Gray Body, Long Wing



Drosophila Melanogaster - Mutant Type
Gray Body, Vestigial Wing

Test Cross

Parents BbVv x bbvv
Gray body longwing Black body vestigial wing
male female

Gametes BV Bv bV bv x bv

			bv
BV	BbVv	-	Gray body, long wing
Bv	Bbvv	-	Gray body, Vestigial wing
bV	bbVv	-	Black body, long wing
bv	bbvv	-	Black body, Vestigial wing

A heterozygous gray body long-winged F$_1$ male crossed to a black body vestigial winged female (test cross).

From the above diagram we can see that, if the two pairs of genes assort at random, we should expect a test cross ratio of 1:1:1:1 i.e. equal numbers of four kinds of offspring. When we made the test cross in this case, however, we did not obtain the expected ratio at all. We find that no gray bodied long winged flies appear nor do we obtain any black bodied vestigial winged flies. Half of the offspring are black bodied, long winged and half are gray bodied vestigial winged. These are just like the original parents with which we started the cross in the beginning.

Looking at the diagram we readily see that no gametes of the BV or bv sorts could have been produced by the F$_1$ male. Only gametes of the types bV and Bv were produced. These gametes are exactly like the gametes that united and produced the F$_1$ male. In other words the genes b and V came into the cross together and stayed together in forming gametes, so also did B and v. Random assortment did not occur. Linkage is an exception to random assortment. The reason for this is the possibility that the genes are on the same chromosome, we should expect that wherever gene b went gene V would also go.

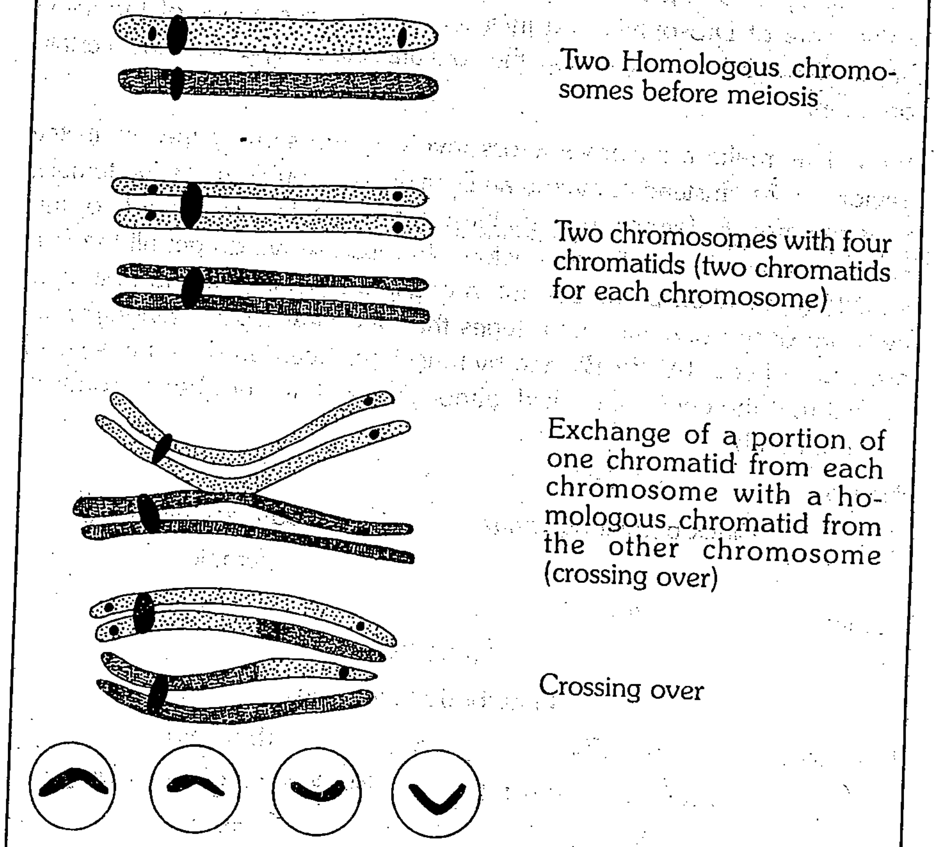
Let us cross on this basis. Assuming the two pairs of genes are on the same chromosome pair, let us write genotype formula in a way which indicates this, so that whereever we see the genotype we will remember that these genes are linked. Instead of writing the formula for the black bodied long winged fly as bbVV, let us put the linked genes in parenthesis thus (bV) (bV). The same genes are present as before, but now they are written in such a way that we can easily remember which two are on the same chromosome.

Now, let us rediagram our cross between a black bodied longwinged fly and gray bodied vestigial winged fly writing the symbols on the assumption that the genes are linked.

Parents (bV) (bV) x (Bv) (Bv)
Black body long wings — Gray body vestigial wing
male — female
Gametes (bV) x (Bv)
F_1 (bV) (Bv)
Test cross
Parents (bV) (Bv) x (bv) (bv)
Gray body long wings — Black body vestigial wings
male — female
Gametes (bV) (Bv) x (bv)
Offspring bv Observed ratio
(bV) (bV) (bv) Black body, long wings 50%
(Bv) (Bv) (bv) Gray Body, vestigial wings 50%

From this, it is easy to see why we get only black bodied long winged flies and gray bodied vestigial winged flies from the test cross. Only two kinds of sperms are formed because the two genes on each chromosome always go together. In this example, linkage is complete. The genes which are located in the same chromosome are linked genes and they form a linkage group. There will be as many linkage groups in each species as the number of chromosome pairs.

Crossing Over


Two Homologous chromosomes before meiosis
Two chromosomes with four chromatids (two chromatids for each chromosome)
Exchange of a portion of one chromatid from each chromosome with a homologous chromatid from the other chromosome (crossing over)
Crossing over

Four Spermatids or one Ootid and three polar biodies.

Note that two of them carry crossover chromosomes and two do not carry crossover chromosomes.

Crossing over

Linkage is not always complete as it is in the previous example. Only in the male of Drosophila and in a few other cases genes of the same chromosome stick completely together, but are able to separate under certain conditions.

Let us make a reciprocal cross involving the same genes as in the previous cross. Instead of mating an F_1 male to a double recessive female, let us mate an F_1 female to a double recessive male. The results of this cross are shown in the diagram below. In this case we do get all the four possible types of offspring, but not in equal numbers. We find that there are more of the original parent types than the new types. Evidently, all four types of eggs bV, Bv, BV and bv must have been produced in spite of the fact that the genes are linked, although they have not been produced in equal numbers.

Parents (bV) (bV) x (Bv) (Bv)
Black body long wings — Gray body vestigial wings
male — female
Gametes (bV) — (Bv)
F_1 (bV) (Bv)
Gray body long wings
Test Cross bV) (Bv) x (bv) (bv)
Gray body long wing — Black body vestigial wing
female — male
Gametes (bV) (Bv) (BV) (bv) x (bv)

Offspring	(bv)		Observed ratio
(bV)	(bV) (bv)	Black body, longwings	42%
(Bv)	(Bv) (bv)	Gray body, vestigial wings	42%
(BV)	(BV) (bv)	Gray body, long wings	8%
(bv)	(bv) (bv)	Black body, vestigial wings	8%

In the diagram, the double line separates the eggs and the offspring which contain the original parental combinations of genes (bV) and (Bv) from those which contain recombinations of these same genes. The parental combinations of genes are present in a majority of the eggs (84%) while the new combinations occur only in 16%. It should appear, then, in the formation of (gametes) most of the eggs the two linked genes stayed together, but that in the formation of a smaller proportion they somehow traded partners. The process which produces the new combination of genes which are linked is known as crossing over. In the above example, the (BV) and (bv) eggs have new combinations. These are cross over gametes. The gametes containing the new combinations are known as CROSSOVER or RECOMBINATION GAMETES. The gametes in which the linked genes remain in their original combinations, here (bV) and (Bv) eggs are called the Non-Crossover Gametes or Parental Type Gametes. Therefore crossing over is an exception to linkage.

Bv bV	}	Non-crossover or parental type of gametes	} 42 + 42 = 84%
BV bv	}	Crossover or recombination type of gametes	} 8 + 8 = 16%

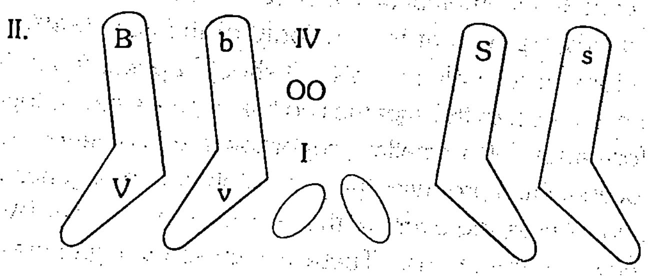
The percentage of crossover = 16

The Cross-over value (COV) = 16%

The percentage of crossover indicates the relative distance between the linked genes. It is customary to express this distance in terms of units, each unit representing one per cent of crossing over. Thus the genes for black body and long wings are 16 units apart on the chromosome. The chromosome map can be drawn as below:

B 16 v b 16 V

The diagram of the chromosomes of Drosophila showing the relationships of the genes for red and sepia eyes and long and vestigial wings.



Linkage is an exception to Mendel's principle of random assortment and occurs because more than one allelic pair of genes is carried on the same pair of chromosomes. Crossing over is in the same way an exception to linkage and occurs because genes may become reassorted though not at random eventhough they are linked, through the exchange of material between homologous chromosomes. Mendel did not discover linkage because he happened to use genes which were on different pairs of chromosomes.

Test for two pairs of genes for linkage:

To find out whether the two pairs of genes are linked or not, let us make a two-pair cross in rabbits. In rabbits a pair of alleles affecting the distribution of coat colour in such a way that the dominant gene 'S' for spotted colour and s for solid colour. Another pair of alleles consists of a dominant gene 'L' for short hair and its recessive allele 'l' for long hair. Let us discover whether or not these two pairs of genes are linked.

Let us make a cross between a spotted short haired rabbit and a solid long haired rabbit and then make a test cross. If the two pairs of genes are not linked, we should expect equal numbers of the four test cross types of offspring. If, however, the genes are linked, there will be more of the parental types and fewer of the new forms.

Let us diagram the cross used for the test, writing the symbols in parenthesis as though the genes were linked even before we know whether they are actually linked or not. In rabbits, as in all higher animals, we find crossing over in both sexes, hence it does not matter whether we use F_1 males or females in the test cross.

The cross is diagrammed as below:

Parents (SL) (SL) x (sl) (sl)
Spotted Solid
Short haired Long haired

Gametes (SL) (sl)

Test Cross: Fl (SL) (sl)
Spotted short haired

(SL) (sl) (sl) (sl)
Spotted Solid
Short haired Long haired

Gametes (Sl) (sl) (Sl) (sL) x (sl)
Non-cross (Cross-
over over)

Offspring		(sl)		Expected ratio If not linked	If linked	Observed ratio
(SL) (sl)	(SL) (sl)	(sl) (sl)	Spotted short haired Solid coloured Long haired	50%	More than 50%	88%
(Sl) (sL)	(Sl) (sL)	(sl) (sl)	Spotted long haired Solid coloured Short haired	50%	Less than 50%	12%

From this diagram we can readily see that if the genes are not linked i.e. if random assortment occurs, we should expect equal numbers of the four kinds of offsprings from the test cross. To put it in other words, we should expect 50% of the test cross offspring to be the parental types and

50% to be of the new combinations. However, if the genes are linked, they will not assort at random and the original arrangement of the genes will tend to hold, changing only where crossing over has occurred, more than 50% of the test cross offspring will be like the parental type and less than 50% will be like the new combinations. In this particular cross, we find 88% of the offspring to be like the parental types and only 12% of them to be of new types. Obviously the genes did not assort at random and they are linked with 12% crossing over. The chromosome map for the two genes are as below:

S 12 L

Modification of the 9:3:3:1 (dihybrid) ratio by linkage:

Linkage modifies the dihybrid ratio (9:3:3:1). Let us illustrate this by taking the example in rabbits.

S = Spotted coloured
s = Solid coloured
L = Short haired
l = Long haired

The genes S and L are linked with a crossover value of 12.

Parents	(SL) (SL) Spotted short haired	x	(sl) (sl) Solid coloured long haired
Gametes	(SL)		(sl)
F_1	(SL) (sl) Spotted short haired		
F_1 x F_1	(SL) (sl)	x	(SL) (sl)

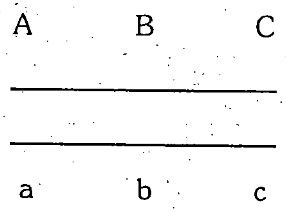
Gametes	(SL) (sl) Non-cross overs 88%	(Sl) (sL) Cross overs 12%	(SL) (sl) Non-cross overs 88%	(Sl) (sL) Cross overs 12%
F_2	(SL) 0.44	(Sl).06	(sL) .06	(sl) .44
(SL) .44	(SL) (SL) 0.1936	(SL) (Sl) 0.0264	(SL) (sL) 0.0264	(SL) (sl) 0.1936
(Sl) .06	(SL) (sl) 0.0264	(Sl) (sl) 0.0036	(sL) (sl) 0.0036	(Sl) (sl) 0.0264
(sL) .06	(SL) (sL) 0.0264	(sL) (Sl) 0.0036	(sL) (sL) 0.0036	(sL) (sl) 0.0264
(sl) .44	(SL)(sl) 0.1936	(sl)(Sl) 0.0264	(sl)(sL) 0.0264	(sl)(sl) 0.1936

S-L- = 0.6936/0.0625 = 11
S-11 = 0.0564/0.0625 = 1
ssL = 0.0564/0.0625 = 1
ssll = 0.1926/0.0625 = 3

The dihybrid ratio of 9:3:3:1 is modified as 11:1:1:3 respectively. Instead of 1/16 of the F_2 individuals being double recessive as seen in random assortment, 3/16 is double recessive because of linkage and 11/16 of the F_2 are the double dominants as against the 9/16 in random assortment.

Theory of Linkage and crossing over:

The theory of linkage and crossing over was proposed by Morgan as early as 1911. Morgan suggested that the genes of the different allelic pairs were arranged in a linear series along the length of the chromosomes and that the amount of crossing over between two pairs of alleles might be related to the distance between them on the chromosome i.e. the farther apart the two pairs of alleles are the greater would be the amount of crossing over.



In the above diagram the exchange of homologous parts of the chromosomes would cause a crossover between pair A, a and B, b only if the exchange took place in the relatively short region between A, a and B, b. On the other hand, an exchange anywhere along the chromosomes would cause a crossing over between pair A, a and pair C, c.

The theory of linear order of genes:

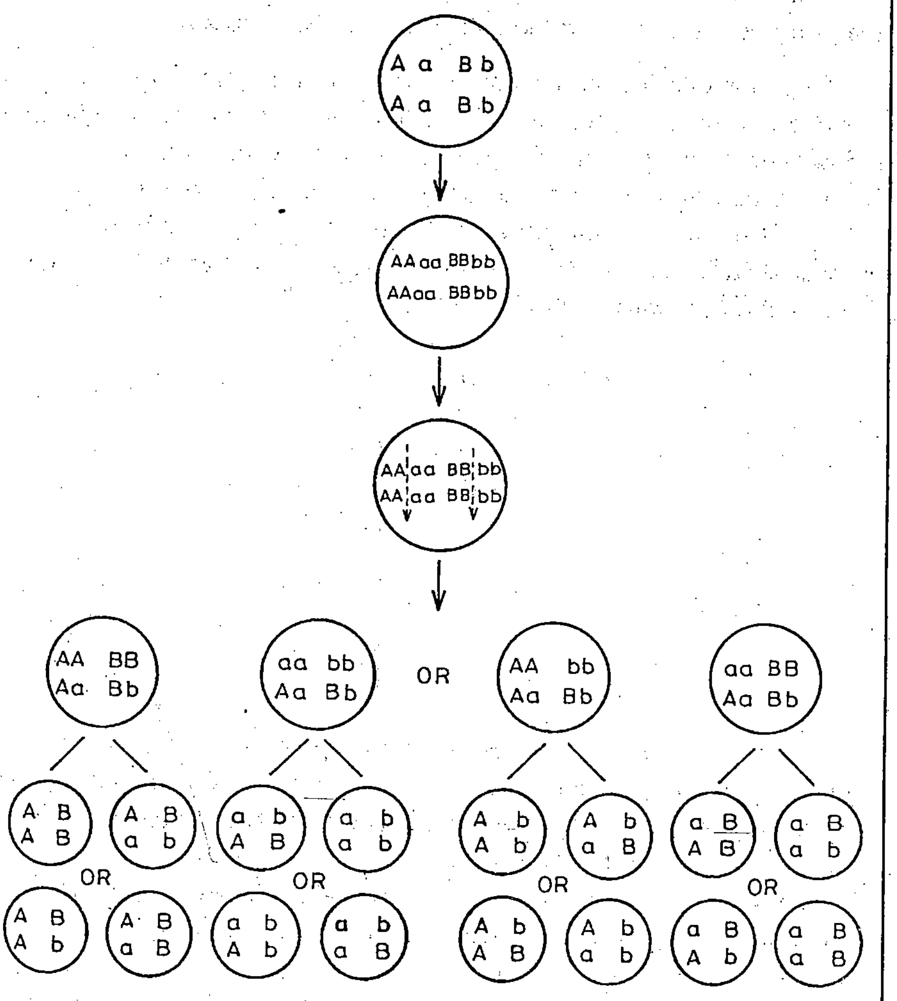
The theory of linear order of genes states that the various alleles carried on any given chromosome are arranged in definite linear sequence and that the different amounts of crossing over between different pairs of alleles is a measure of the relative distances between their positions on the chromosomes. The position occupied by one or the other member of a pair of alleles is called the locus of that pair (loci is plural).

Let us illustrate this principle with an example.

In Drosphila: The following three factors are known:

W = Red eyes
w = White eyes
Y1 = Gray body
y1 = Yellow body
Bi = Normal wings
bi = Bifid wings

When crosses are made involving factors for white eye and yellow body (w and yl) it is found that these genes are linked with 1.5% crossing over


A a B b
A a B b
AA aa BB bb
AA aa BB bb
AA aa BB bb
AA aa BB bb
AA BB
Aa Bb
aa bb
Aa Bb
OR
AA bb
Aa Bb
aa BB
Aa Bb
A B
A B
A B
a b
OR
A B
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OR
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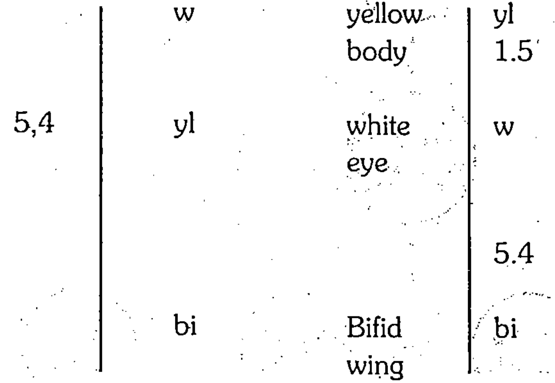
a B
a B

Dance of Chromosomes

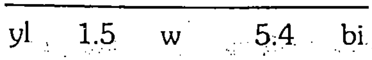
The gamete formation with the new combinations of chromosomes due to chance orientation of bivalents at the equatorial plate and the crossing over between homologous chromosomes.

When crosses are made involving factors for white eye and bifid wings (w and bi) they are found to be linked with 5.4% crossover.

These three factors namely w, yl and bi must therefore be located on the same pair of chromosomes. The crossover value between white and bifid is nearly 4 times than the crossover value between white and yellow. This might be taken to indicate that the factor for bifid is about 4 times as far from the factor for white eye as is the factor for yellow body. If the genes are on a line on the chromosome it is possible for them to be arranged in one of the two ways as shown below:



To determine which of these relationships are correct, we have to determine the crossover percentage between yellow and bifid. If the genes are in the order, w, y1 and bi, then the crossover between yellow and bifid is 5.4 - 1.5 = 3.9. If they are arranged as yl, w and bi. The crossover value between yellow and bifid is 5.4 + 1.5 = 6.9. When crosses are made between yellow and bifid they give a crossover value of 6.9, showing yl is on one side of the factor and bi is on the other side of the factor w. The factors yl, w and bi are arranged in a straight line and the chromosome map is as follows:

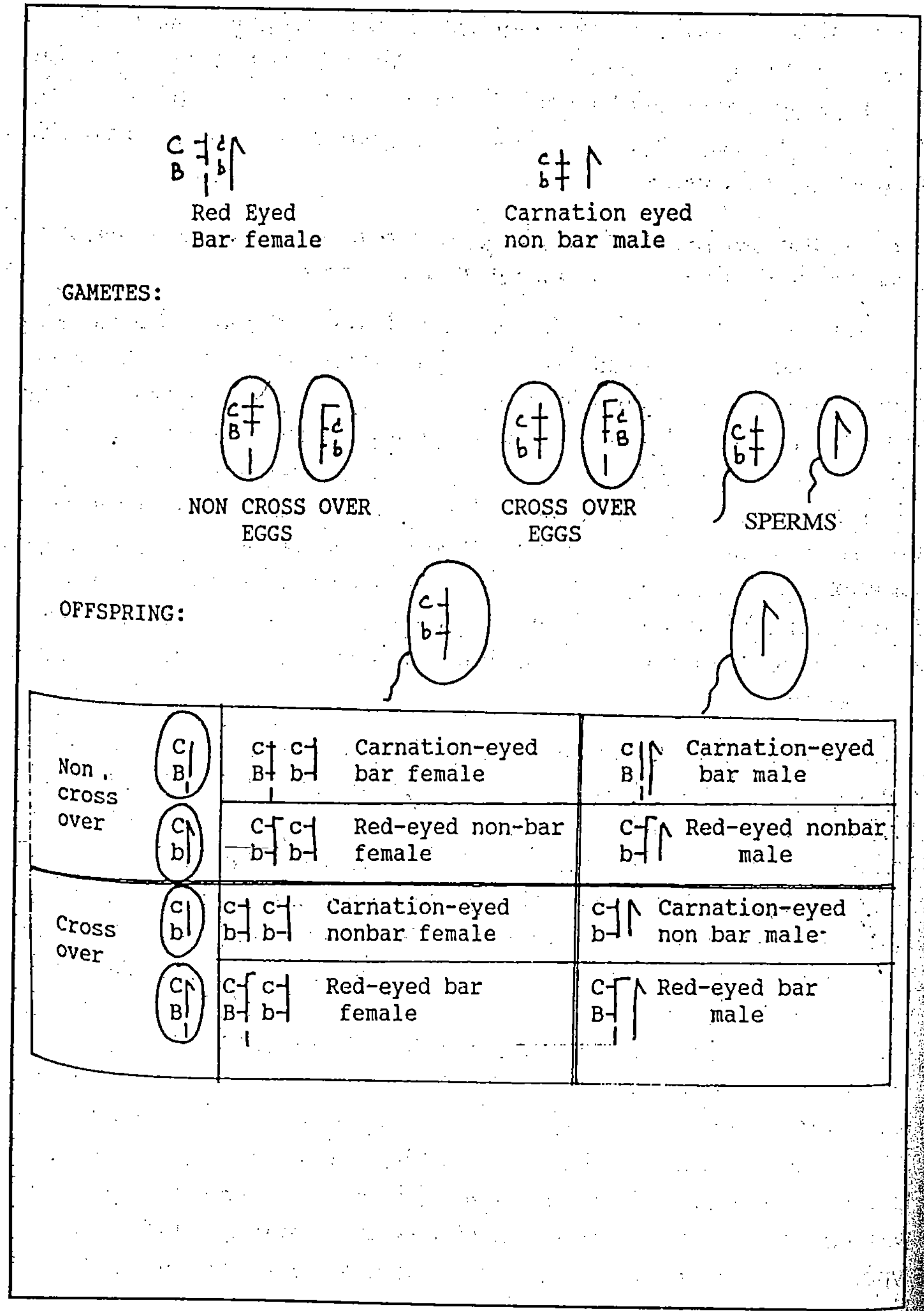


To put it in general terms, if in regard to three linked genes A, B and C, A crossover from B with a certain percentage and B crossover from C with a certain percentage then the crossover percentage between A and C will be either the sum or the difference of these two percentages. Since these relationship are also of points on a line, we have strong indication that the genes are located in a definite linear order along the chromosome, the crossover percentage being more or less proportional to the distances between their locations.

The cytological proof of crossing over between chromosomes of a pair and the concurrent crossing over of genes:

An essential part of the linear order theory is the idea that genetic crossing over is the result of the physical exchange of parts between homologous chromosomes (cytological crossing over). The genetic data from the linkage studies left no doubt that these exchanges actually occur, eventhough it had been entirely impossible to observe them cytologically, since there is no visible difference between the two chromosomes of a homologous pair. In order to be able to see whether or not an exchange takes place, it is necessary that the two synapsing chromosomes differ visibly in two different regions. Then, if exchange does occur the crossover chromosomes (chromatids) will be distinguishably different from the original chromosomes.

Let us illustrate the above by taking a specific example. Stern (1931) discovered a strain of Drosophila in which a portion of the Y chromosome had broken off and became attached to the end of one of the X chromosomes. This X chromosome with the piece of Y attached could be readily recognised under the microscope. Stern also discovered another strain of Drosophila in which one of the X chromosomes was broken into two. By suitable crossing over, Stern was able to produce female flies in which one of the X chromosomes had the fragment of Y attached and the other X chromosome was broken into two. The broken X chromosome carried the dominant gene B for bar eye in which the eye becomes much narrowed and the recessive gene c for carnation eye, a colour gene. The X chromosome with the piece of Y attached carried the recessive gene b for non-bar eye and the dominant gene C for red eye. When such a female was bred to a non-bar carnation eyed male, the offspring would be of four types, two of which would be non-crossovers and two crossovers as illustrated in the diagram:


Red Eyed
Bar female
Carnation eyed
non bar male
GAMETES:
NON CROSS OVER
EGGS
CROSS OVER
EGGS
SPERMS
OFFSPRING:
Non cross over
Carnation-eyed bar female
Carnation-eyed bar male
Red-eyed non-bar female
Red-eyed nonbar male
Cross over
Carnation-eyed nonbar female
Carnation-eyed non bar male
Red-eyed bar female
Red-eyed bar male

The chromosomes of the female offsprings were studied. If crossing over of genes is accompanied by exchange of parts of chromosomes, it should be possible to see the results of the chromosomal exchange in the crossover offspring. The cytological results showed definitely that crossing over of genes was accompanied by chromosomal exchange. In the female offspring, one X chromosome always came from the father and should therefore be normal. The other came from the mother and should be broken into two or should have a piece of the Y attached.

In the non-crossover females, this was true. In the crossover females, however, it was clear that the X chromosome from the mother was the result of a chromosomal crossover. In the carnation eyed non-bar female offspring, the maternal X chromosome was apparently normal, neither broken nor carrying a piece of Y chromosome. In the red eyed bar offspring, the maternal X chromosome was not only broken, but one of the broken piece carried the piece of the Y chromosome. This is a clear proof that genetic crossing over is accompanied by and dependent upon cytological crossing over and that there is an actual exchange of material between a pair of homologous chromosomes.

There is no doubt, then, that linkage and crossing over have a definite cytological basis. This provides more parallel between the behaviour of chromosomes and behaviour of genes.

Certain genes do not assort at random but occurred in paired groups (linkage groups) which tend to be transmitted as units. The chromatin material also gathered into paired (chromosomes) which tend to be transmitted as units.

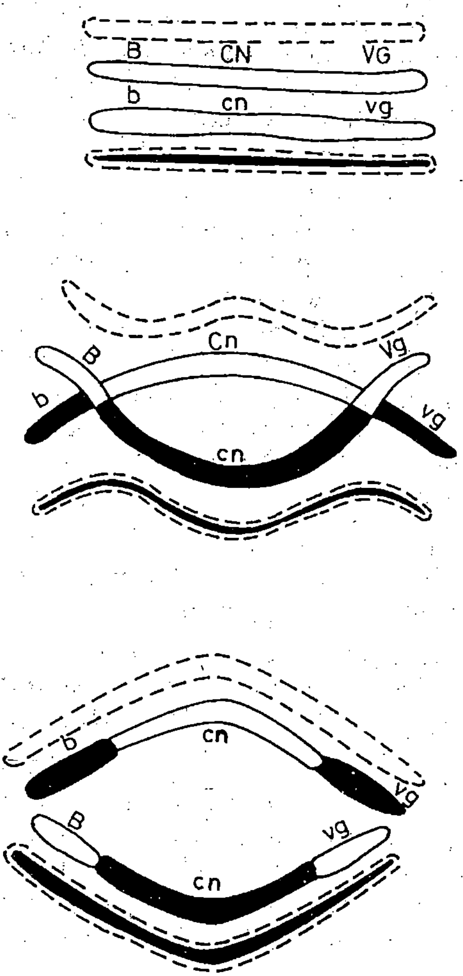
The genes of linkage group do not stay completely together as a rule, but during maturation of homologous parts of the paired groups of genes are exchanged with a definite frequency (genetic crossing over). The paired chromosomes also exchange homologous parts of their lengths during the maturation of germ cells (cytological crossing over).

When a crossing over occur at one point, another crossing over does not occur within a certain distance of it. This phenomenon is known as interference when crossing over occurs at two places, it is called as double crossing over.

The mapping of chromosomes:

The discovery of linkage, linear order of genes and measurement of

Double Crossing Over



Three genes on the chromosome from Drosophila which are linked - wild type alleles on one chromatid and black, cinnabar and vestigial alleles on another chromatid. Double crossing over affects linkage.

their relative distances lead to the construction of chromosome map. The chromosome map indicates the sequence and relative distances between genes on the chromosomes. It is the figurative expression of the chromosome with the genes measured on it in their linear order and relative positions based on the crossover units. Sturtewant in 1913, first constructed a map showing the relative positions of five different loci on the x chromosome of Drosophila together with approximate distances of each other. In the following years, more and more mutants were discovered, their linkage relations were studied and by 1925 it was possible to construct the detailed map of all chromosomes of Drosophila.

Let us examine a three point linkage test in which individuals heterozygous for each of three linked pair of alleles are mated to the triple recessive homozygotes.

In Drosophila the following three pairs of alleles are carried on the second chromosome:

B = Gray body
b = Black body
Cn = Red eye
cn = Cinnabar eye
Vg = Long wing
vg = Vestigial wing

The genes b and vg are linked with 17% crossover value. The genes cn and vg are linked with a crossover value of 9.5%. The genes b and cn are also linked with a crossover value of 9.0%. This indicates the order of the genes on the second chromosome as follows:

---

b 9.0 cn 9.5 vg

The expected crossover value between b and vg as per the additive rule 9.0 + 9.5 = 18.5. The observed double crossover between b and vg = 17.00. The discrepancy between expected and observed = 18.5 - 17.0 = 1.5. The discrepancy of 1.5 is double the value of the double crossover namely 0.75 i.e. twice the value of double crossover have not been included in the expected crossover value. Therefore, crossover value between b and vg = 9.0 + 9.5 - 2 x 0.75 + 18.5 - 1.5 = 17.0.

The observed crossover value which is low denotes that the crossovers between b and cn and cn and vg are not independent events. The expected double crossovers between b and cn and cn and vg based on independent events is equal to 0.09 x 0.095 = 0.0085 = 0.85%. But actually only 0.75% of the double crossover has occurred. This only shows the crossover in these two regions do not occur independently. Moreover the crossover occurring at one point prevents a second crossover within a certain distance of it. This phenomenon is called interference. The effects of interference is expressed in terms of coincidence. Coincidence is the ratio of the observed number of double crossovers to the expected numbers (or the ratio of the observed percentage of double crossovers to the expected percentage of double crossovers).

The coincidence of double crossover in the above example is equal to 0.75/0.85 = 0.88 or 88%. This means only 88% of the expected double crossovers have actually occurred i.e. there is very little interference between crossovers in the two regions to which the data relates.

Coincidence may have a range of value from 0 to 1. If the value is 0, the interference is complete. This means the genes are located very close together. If the value is 1, there is no interference. This means the genes are located farther apart.

In Drosophila, the degree of interference is more, if the genes are located at 10 units apart and less. It diminishes as the genes concerned are 10 units apart and more. The degree of interference is practically not seen between genes located 40 units apart or more than that.

Parents Bb, Cn cn, Vg vg

Bb Cn Vg vg x bb cn cn vg vg

(B Cn Vg) (b cn vg) x (b cn vg) (b cn vg)

The gametes produced by the female and their members are as follows:

PHENOTYPES	Chromosome received from the mother	No.	% of Total	Gene combination
Gray body, red eye, long wing	B Cn Vg	332	82.25	non-cross-over parental
Black body, cinnabar eye, vestigial wing	b cn vg	326		
Gray body, red eye vestigial wing	B Cn vg	34	8.75	Single cross-over between cn and vg loci only
Black body, cinnabar eye, long wing	b cn Vg	36		
Gray body, cinnabar eye, vestigial wing	B cn vg	31	8.25	Single cross-over between b and cn loci only
Black body, red eye, long wing	b Cn Vg	35		
Gray body, cinnabar, long wing (recombination	B cn Vg	4	0.75	Double crossover between both cn, vg and b, cn loci
Black body, red eye, vestigial wing	b Cn vg	2		
		800		

Total number of single crossovers } 34 + 36 + 4 + 2
between cn and vg loci } = 76
Total number of single crossovers } 31 + 35 + 4 + 2
between b and cn loci = 72
Percentage of crossovers between } 76/800 x 100
cn and vg loci } = 9.5
Percentage of crossovers between } 72/800 x 100
b and cn loci } = 9.0

The chromosome map is as below:

B 9.0 cn 9.5 vg

Linkage groups:

By studying the crossover percentages, genes may be located in their relative positions along the chromosomes. By considering each per cent of crossing over as a unit of distance, we may make maps of chromosomes of any species in which we know a sufficient number of genes. A series of gene pairs which are linked with other is considered to be on the same pair of chromosomes and such a series of gene pairs is known as linkage group. The members of a linkage group show linkage with each other but they show random assortment with regard to the genes of any other linkage group. Moreover the number of linkage groups should be equal to the number of pairs of chromosomes. In Drosophila melanogaster, there are four pairs of chromosomes and four linkage groups are known. Of these, one is known as sex linkage which contains the genes located on the first or sex chromosome (sex linked genes). The second and third linkage groups are large and are carried on the large second and third chromosomes respectively. The fourth linkage group is very small as it contains about five genes only.

In corn, there are ten pairs of chromosomes and ten linkage groups are known. In garden peas there are seven pairs and seven linkage groups are known.

The number of linkage groups in given species is equal to the number of pairs of chromosomes. It can never be more than the number of pairs of chromosomes. The number of linkage groups may be less than the number of pairs of chromosomes in which case some linkage groups might not have been discovered.

Under given conditions, the crossover value between any two pairs of genes is always constant. The following conditions may influence the crossover value:

1. Age
2. Temperature
3. Radiation
4. Arrangement of a part of a chromosome
5. Arrangement of genes in a double heterozygote.

We have learnt in this chapter that corresponding to each chromosome there is a group of linked genes which occur in a definite linear order in the chromosome and that maps of the chromosomes showing the location of genes may be constructed. From this, there emerge new paralleles between the behaviour of genes and the behaviour of chromosomes. They are:

1. At the time when genetic crossing over takes place, the genes are arranged in a linear order. At the time when cytological crossing over takes place, the chromatin material is in an attenuated linear arrangement.
2. The number of pairs of chromosomes is, in general, definite and constant for any given species. The number of linkage groups in those species which have so far been studied is equal to or at least never exceeds the number of pairs of chromosomes of the species.

EXERCISE

1. Linkage is an exception to random assortment. Prove this concept.
2. Crossing over is an exception to linkage. Prove with example in Drosophila.
3. Give the cytological proof of crossing over.
4. Write short notes on
   a) Theory of linkage
   b) Theory of linear order of genes
   c) Chromosome map
   d) Linkage groups
   e) Modification of dihybrid ratio due to linkage
5. A pair of genes in poultry results in R (rose comb) and its recessive allele r (single comb). Another pair of alleles result in G (non-frayed) and g (frayed). A homozygous non-frayed hen with single comb was mated to a frayed homozygous rose comb rooster. The F_1 were crossed to frayed single combed fowls. The results are as follows:

Non-frayed rose comb	58	Frayed single comb	56
Frayed rose comb	51	Non-frayed single comb	55

Are these genes G, g and R, r linked?

6. A fully heterozygous gray bodied (B) normal winged (Vg) female Drosophila was crossed with a black bodied (b) vestigial winged (vg) male gave the following results:

Grey normal	126	Gray vestigial	24
Black vestigial	124	Black normal	26

Are these genes, B, b and Vg, vg are linked? If so what is the percentage of crossing over?

7. A homozygous vestigial (vg) stock was crossed with a homozygous Cinnabar (Cn) stock. The F_1 females were back crossed to males homozygous for cn and vg. The following phenotypic classes were obtained:

Cinnabar	171	Vestigial	181
Cinnabar-vestigial	38	Wild type	30

Assume linkage and calculate the linkage between cn and vg loci.

8. In poultry four pairs of genes are linked. F—brizzled feathers; f—plain feathers; H—crested head; h—non-crested; I—white; i—coloured; G—non-frayed feathers; g—frayed feathers. The crossover value between the genes are as follows: H and I-18% G and H-30% I and F-18% G and I-36% H and F-27%. In what order do these genes occur on the chromosome?

9. In Drosophila white eye (w) miniature wing (m) and forked bristles (f), the sex-linked and recessive to the wild type characters red eyes, long wings, and straight bristles. In a cross of wfm x WFM the F_1 females crossed with wfm males gave the following:

	Per cent
White, forked, miniature	26.8
Red, straight, long	26.8
White, straight long	13.2
Red, forked, miniature	13.2
White, straight, miniature	6.7
Red, forked, long	6.7
White, forked, long	3.3
Red, straight, miniature	3.3

Designate non-cross over, single crossover and double crossover classes. Determine the percentage of crossing over between white and forked, white and miniature, and miniature and forked, and from this determine the order of genes in the chromosomes.

10. In Chinese primroses short style is dominant over long, megenta flower over red, and green stigma over red. When from the cross of homozygous short, magenta flower, green stigma by long, red flower, red stigma, the F_1 was crossed with long red flower red stigma the following offspring were obtained:

Style	Flower	Stigma	Number
Short	magenta	green	1063
Long	red	red	1032
Short	magenta	red	634
Long	red	green	526
Short	red	red	156
Long	magenta	green	180
Short	red	green	39
Long	magenta	red	54

Map the chromosome in which these genes line.

कृपया रेषा मारून
पुस्तक खराब करू नये.

Chapter 16

Mutliple Alleles

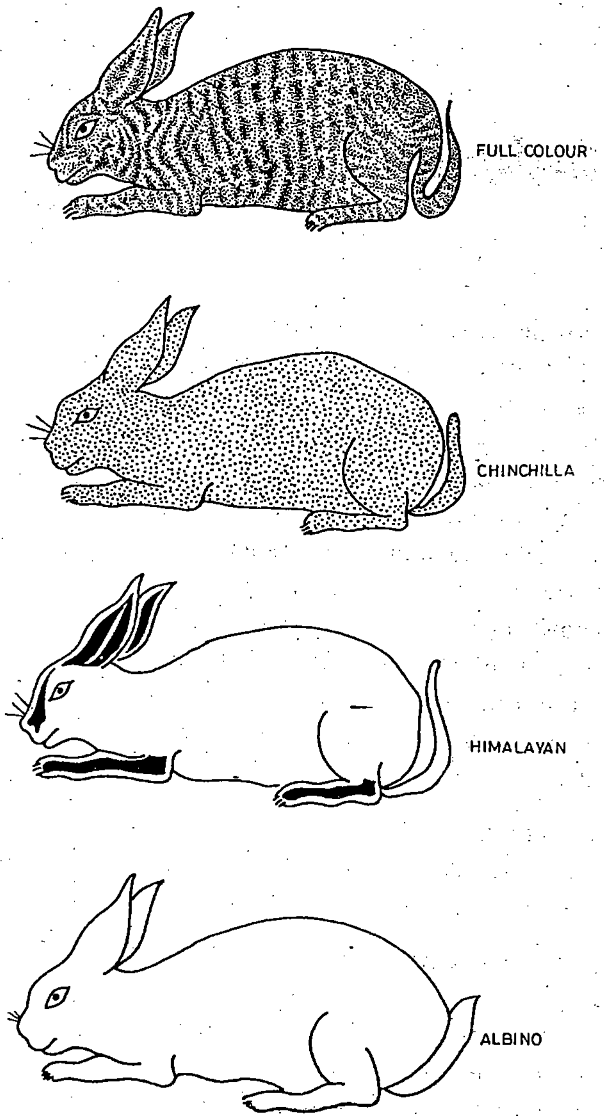
The term alleles apply to the members of a pair of contrasting genes. In speaking of alleles, we have so far considered them occurring in pairs. Cases are known where sets of alleles contain three, four and even twenty or more alleles. Such sets of alleles are called MULTIPLE ALLELES. No matter how many members a set of alleles may contain, only two of them may normally occur in a somatic cell and only one in a gamete. Therefore, in case of multiple alleles also one individual may either be homozygous or heterozygous genotypically. However, several forms of heterozygotes can occur.

We have already learnt how to recognize the fact that two factors are alleles. Let us now see how we could recognize the presence of a third allele.

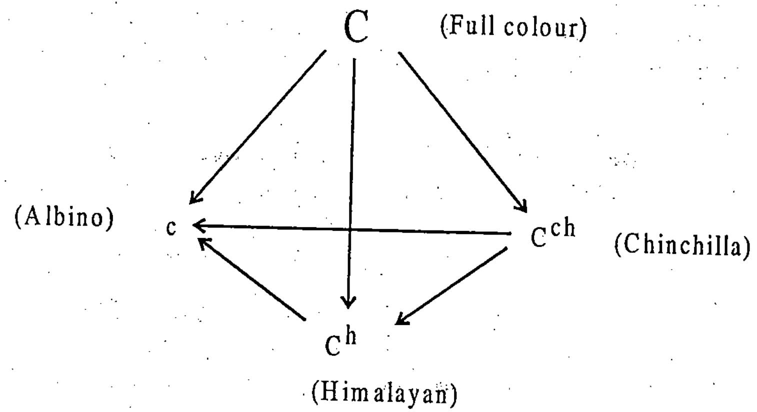
If we cross a homozygous coloured rabbit with an albino, we obtain an F_1 generation in which all the individuals are coloured. The F_2 generation from this cross consists of both coloured and albino rabbits in the ratio of 3:1. We know from this, that colour is dependent upon a dominant gene,

Multiple Allele Inheritance

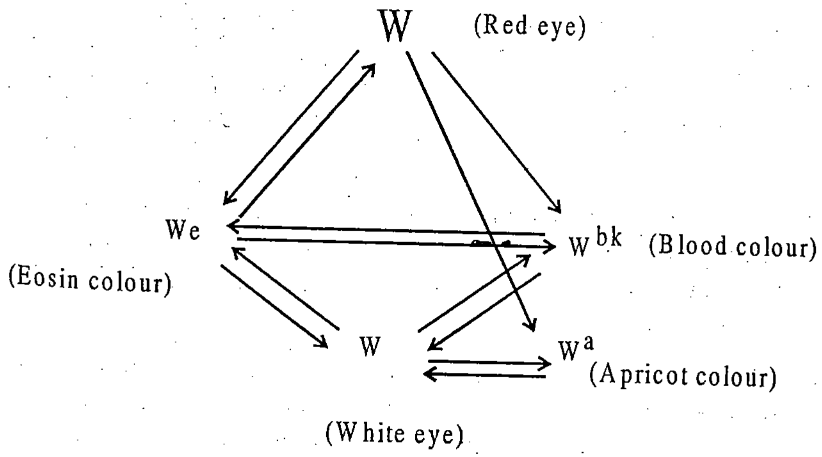
(Coat Colour in Rabbits)


FULL COLOUR
CHINCHILLA
HIMALAYAN
ALBINO

The Direction of Mutation


C
(Full colour)
(Albino)
c
C^{ch}
(Chinchilla)
C^h
(Himalayan)

Skin Colour in Rabbits


W
(Red eye)
We
(Eosin colour)
W^{bk}
(Blood colour)
W
W^a
(Apricot colour)
(White eye)

Eye Colour in Drosophila

albinism upon a recessive gene and the two are alleles.

In addition to complete colour, however, we have known a condition in which only the extremities are coloured, the rest of the fur being white. This condition is known as the Himalayan. These rabbits have pigmented ears, nose, tips of the feet and tip of the tail. The Himalayan condition is a partial colour, obviously different from the full colour of a completely gray or black rabbit.

If we cross a full coloured rabbit with a Himalayan, we obtain F_1 all of which are full coloured. The F_2 generation consists of full coloured rabbits and Himalayan rabbits in the ratio of 3:1. Obviously full colour is dependent upon a dominant gene and Himalayan upon its recessive allele.

What could be expected if we cross a Himalayan and albino rabbit? If the Himalayan and the albino mutations represent changes in different genes, we should expect two pairs of genes to be involved in the cross. In this case, we should obtain a two-pair ratio in the F_2 just as we found to be the case when albino rat was crossed with a black one. When the cross is made between the Himalayan rabbit and an albino, the F_1 are all Himalayans and not full coloured as would be expected if two pairs of genes were concerned. The F_2 does not give a two-pair ratio, but consists of Himalayans and albinos in the ratio of 3:1. We must conclude, therefore that the genes for Himalayan and albinism are alleles. But if the full colour and the albino genes are allelic and the full colour and Himalayan genes are allelic and finally the genes for Himalayan and albinism are allelic, it is evident that all the three genes must belong to the same set of alleles. They form a multiple allelic series.

A gene always occupies a particular locus on a chromosome and it is evident that any individual rabbit could carry only two of these three genes at a time and any gamete could carry only one. This is found to be the case in breeding experiments.

If we designate 'C' as the gene for full colour and 'c' as the gene for albinism, we may designate the third allele in this series c^h. The superscript h stands for Himalayan. It is customary in designating multiple alleles to give the one which is dominant to all the others a capital letter, the one recessive to all the others the corresponding small letter and the alleles

which are in between the small letter with an appropriate superscript. Thus, in this case, colour is dominant to both Himalayan and albinism. We, therefore, designate it C. Himalayan is recessive to colour, but dominant to albinism. We designate it as c^h. Albinism being recessive to both the others, receives the designation c.

Rabbits could then be of the following genotypes in regard to this particular set of genes:

CC }
CC^h }
Cc } Full colour

c^hc^h }
c^hc } Himalayan

cc } Albino

Still another allele has been found in this series. These rabbits known as Chinchillas, in which all traces of yellow are lacking, giving it a characteristic silvery gray appearance. When homozygous Chinchillas are crossed with full coloured rabbits or with Himalayan or with albinos, the F_2 always show a one-pair ratio, indicating that the gene for Chinchilla is allelic to the other three. It is recessive to full colour, but dominant to other two, so that we designate Chinchilla as c^{ch}. Taking all these alleles into consideration, a rabbit could carry any two of them and a gamete could contain but one of them so that rabbits could be of the following genotype in regard to these genes:

CC }		$C^{ch}c^{ch}$ }	
CC^{ch} }		$c^{ch}c^h$ }	Chinchilla
Cc }	Full colour	$c^{ch}c$ }	
Cc^h }			
c^hc^h }			
c^hc }	Himalayan	cc	- Albinism

Let us consider how this state of affairs came about. The original wild rabbits from which the same rabbits have been domesticated were gray and must have carried the genes CC. Somewhere, at some time, by mutation, the chemical composition of one of the C genes in a developing germ cell became changed in such a way as to be unable to function in the production of colour. Let us call the changed gene c.

The gamete containing c in place of C would probably unite with one containing the usual C, since mutations are relatively rare. The resulting individual would be of the formula Cc and would show no sign that its genotype was in any way different from other rabbits. It would, however, produce germ cells of two kinds, half of them containing C and half c. Eventually, in a later generation two gametes containing c might unite, thus producing an individual of the formula cc, which would be an albino. The phenotypic appearance of a mutation thus occurs much later than the mutation itself, if the mutation is a recessive autosomal one. A dominant mutation would show up in the first generation. Even a recessive mutation, if it were sex linked could show up in the first generation in the heterogametic sex.

Similarly, at some time the gene C would have changed to c^h in a developing germ cell resulting in a Himalayan. A rabbit could have received the gene c from each parent and the Himalayan individual c^hc^h results. Likewise the change in the composition of the gene C to c^{ch} results in an individual c^{ch} c^{ch} which is the Chinchilla. In this way, numerous series of multiple alleles have been formed in various animals and plants and in man.

Example in Drosophila:

Another example of multiple allelic series could be given in the eye colour of Drosophila. In addition to the sex linked gene red eyes and white eyes, we have studied, this set has many members including not only red and white, but wine, coral, blood, eosin, cherry, apricot, buff, tinged and ivory.

The multiple alleles for the eye colour in Drosophila may be designated as follows:

W - red
w^w - wine

w-white
w^{co} - coral

w^b - blood
w^a - apricot
w^{bf} - buff
w^h - honey
w^p - pearl

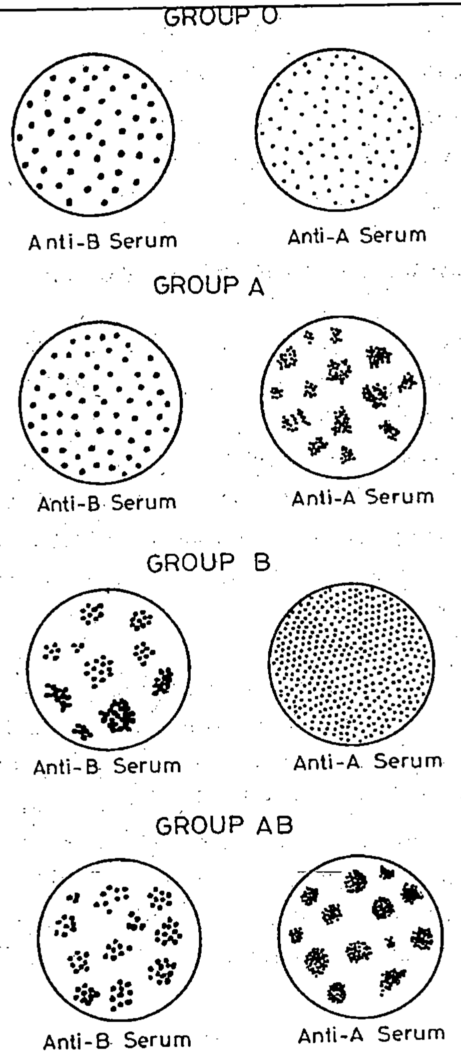
w^c - cherry
W^e - eosin
w^t - tinged
w^{ee} - ecru
w^i - ivory

Example in human beings - ABO Blood groups:

In man, the blood groups are inherited on the basis of multiple alleles. In 1900 Kari Landesteiner, working in a Medical Laboratory in Vienna, discovered that when the RBCs of one person were mixed with the blood serum of another person agglutination sometimes occurred. The reaction did not happen in every combination of serum and cells, but only when the red cells of certain people were mixed with the serum of certain other people. Obviously, normal antibodies must occur in some bloods and normal antigens must occur in the red cells of some people.

There are two antigens in human red cells and two corresponding antibodies in the serum. The antigens were named A and B. A person might have one of these antigens in his red cells, or he might have the other, or both or neither. There are thus four kinds of persons in the world in regard to the two antigens and these four possibilities are known as the blood groups. A person having antigen A in his cells is said to belong to group A; a person having antigen B belongs to group B; a person having both antigens is group AB; and a person having neither antigen belongs to group O.

Whatever antigen a person has in his cells, the corresponding antibody is lacking in the serum. This obviously must be so, since a person of group A had the antibody against A in his serum he would agglutinate his own cells (isoagglutination) and die. When an antigen is not present in the cells, the corresponding antibody is present. Thus a person of group A has antigen A, but no antibody against A; he has no antigen B but does have the antibody against B. The antigens in the cells and the antibodies in the serums


GROUP O
Anti-B Serum
Anti-A Serum
GROUP A
Anti-B Serum
Anti-A Serum
GROUP B
Anti-B Serum
Anti-A Serum
GROUP AB
Anti-B Serum
Anti-A Serum

AB	Both A and B antigens. Agglutination with both Anti-A and Anti-B serums.
A	Agglutination with Anti-A serum
B	Agglutination with Anti-B serum
O	No antigens. No agglutination

of all four groups are as below:

Blood group	Antigens in cells (RBC)	Antibodies in serum
0	-	anti A and anti B
A	A	anti B
B	B	anti A
AB	A, B	Nil

The inheritance is explained on the basis of multiple alleles.

I^A represent the gene for antigen A

I^B represent the gene for antigen B

i represent the gene for no antigen

I^A and I^B are each dominant to i. But neither is dominant to the other. I^A and I^B are called co-dominant alleles and when both are present in an individual both antigens are produced. The blood type and its corresponding genotype is given below:

Blood group (Type)	Genotype
O	ii
A	I^AI^A, I^Ai
B	I^BI^B I^Bi
AB	I^AI^B

The inheritance of blood groups, based on the study of thousands of families is given in the following table.

Blood groups of parents	Blood groups which may occur in children	Blood groups which do not occur in children
OXO	O	A, B, AB
OXA	O, A	B, AB
AXA	O, A	B, AB
OXB	O, B	A, AB
BXB	O, B	A, AB
AXB	O, A, B, AB	-
OXAB	A, B	O, AB
AXAB	A, B, AB	O
BXAB	A, B, AB	O
ABXAB	A, B, AB	O

It is seen from the above table that antigen A never appears in a child's blood unless it was present in the blood of at least one of the parents. Likewise, antigen B never appears in a child's blood unless it was present in the blood of the father or mother or both.

The inheritance of blood groups is of great value in the practical applications in the field of legal medicine. The medico-legal application of the blood groups are given in the table as follows:

Blood Transfusion Compatibilites

Blood Group	**Antigens Present**	**Antibodies Present**	**Red Cell Types Agglutinated**	**Transfusions Accepted From**
A	A	Anti-B	B, AB	A or O
B	B	Anti-A	A, AB	B or O
AB	A and B	None	None	A, B, AB or O
O	None	Anti A and Anti B	A, B, and B	O

O - Universal donor.

AB - Universal recipient

Blood group of child	Blood group of mother	Blood group to which the father cannot belong
O	O	AB
O	A	AB
O	B	AB
A	O	O, B
A	B	O, B
B	O	O, A
B	A	O, A
AB	A	O, A
AB	B	O, B
AB	AB	B

The practical importance of the blood group was in connection with blood transfusion. A test is made before every transmission, to make sure that the blood given will not have its cells agglutinated by the serum of the patient.

Recipient (Agglutinins in serum)	Red cells of Donor			
	AB	A	B	O
AB + No agglutinins (O)	-	-	-	-
A + agglutinin (B)	+	-	+	-
B + agglutinin (A)	+	+	-	-
O + agglutinin (A, B)	+	+	+	-

+ = agglutination of cells - = no agglutination

The Secretor trait:

The blood group antigens A and B are found in two chemical forms. The secretors have water soluble antigens, and therefore, some of these pass out into body secretions like discharges from the eye, nose, saliva. The non-secretors have antigens which are soluble only in alcohol. Thus the secretors can be identified by tests on blood as well as on the body secretions. The ability to secrete these antigens (secretor) is due to a dominant gene, with the recessive allele, s, representing the non-secretor condition. Group O secretors secrete H substance only. Secretors of the other groups secrete not only their group-specific substance A, B or AB but H substance as well.

The M-N Blood types:

The M and N antigens were discovered in 1927. Unlike antigens A and B, no natural antibodies against them are found in human serums. Moreover, they practically never provoke the formation of antibodies against themselves when one or the other of them gets into the blood of a person lacking it. They are therefore, of no practical consequence in transfusions.

Antiserum (serum containing antibodies) with which these antigens will react must be artificially produced by injecting the antigens into rabbits. By the use of anti-M and anti-N serums produced in this way, it has been found every individual possesses either antigen M, antigen N, or both M and N, so that every member of the human species can be classified as belonging to one of three types M, N or MN. Genetic studies of large number of families have shown that two M type parents produce only M type children, two N type parents have only N type children; when one parent is type M and the other N, the children are invariably MN; and when both parents are MN, all three types of children occur in the 1:2:1 ratio. These observations lead us to conclude that a single pair of alleles M and N are responsible for the production of M and N antigens respectively and that neither allele is dominant to the other (co-dominant alleles).

THE MN BLOOD TYPES

M-N type	Genotype	Reactions with antiserum	
		Anti-M	Anti-N
M	MM	+	-
N	NN	-	+
MN	MN	+	+

THE INHERITANCE OF THE ANTIGENS M AND N

Antigen type of parents	Antigen types which may occur in children	Antigen types which do not occur in children
M x M	M	N, MN
M x N	MN	M, N
N x N	N	M, MN
M x MN	M, MN	N
N x MN	N, MN	M
MN x MN	M, N, MN	-

MEDICO-LEGAL APPLICATIONS OF THE ANTIGENS M & N

Antigen type of child	Antigen type of mother	Antigen type to which father cannot belong
M	M	N
M	MN	N
N	N	M
N	MN	M
MN	M	M
MN	N	N

The Rh Blood antigens:

The Rh antigens, like those of the M-N systems, differ from A and B antigens in that normal antibodies against them are not found in human serums. They also differ from M-N antigens, however, because the introduction of an Rh antigen into the blood which does not already possess it does induce antibody production. They, thus become important in blood transfusion.

The original Rh antigen was discovered by Landsteiner and Weiner, who injected the blood cells of Rhesus monkeys into rabbits. The rabbits produced an antibody against an antigen of monkey cells. When the antiserum, thus prepared was tested against human blood, it agglutinated the red cells of about 85% of persons tested but failed to react with the cells of the remaining 15%. The human antigen identified in this way was called Rh, from the first two letters of Rhesus. Those with antigens were designated as Rh positive, those without it Rh negative.

The difference between Rh positive and Rh negative depended upon a single pair of genes, with the gene responsible for the Rh positive condition being dominant. The blood type and genotypes are as follows:

Rh type	Reactions with Rho antiserum	Genotype
Positive	+	RR, Rr
Negative	-	rr

The multiple allelic series in the Wiener and Fisher-Race system is as follows:

GENE SYMBOLS		
Wiener	Fisher-Race	Type
R^0	cDef	Rh positive
R'	CDeF	"
R"	cDEF	"
R^2	CDEF	"
r'	CdeF	"
r"	cdEF	"
r^Y	CdEF	"
r	cdef	Rh Negative

Weiner, an American Geneticist proposed that there were eight main alleles at locus for this characteristic on the chromosomes. This was possible because some of the alleles produced two or three of the positive antigens.

R.A. Fisher, an English Geneticist proposed a theory called pseudo allelism to explain the relationship. This hypothesis holds that these genes are so close together on the chromosome that they ordinarily move and act as one gene. Such genes are called pseudo alleles. The dominant alleles CDE results in Rh positive condition and the recessive alleles cde results in

Rh negative condition. Recently, one more set of genes F, f are also found to be associated so that the presence of any one of the dominant gene results in Rh positive type and recessive alleles namely cdef results in Rh negative condition.

The practical significance of Rh blood types:

If an Rh negative person receives blood from an Rh positive donor, there will be no reaction, if the person has never had a positive transfusion before, since there are no maternal antibodies to Rh antigens. But the presence of Rh antigen in the recipient will stimulate the production of Rh antibodies. If the same person receives another transfusion from the same donor at a latter date, this time the cells of the donor will clump as they react with antibodies now present, and the recipient may die. Hence, no Rh negative person should ever be given a transfusion from an Rh positive person, eventhough it may be the first transfusion.

The Rh factor has great significance in child birth. It has been found that an Rh negative woman who has been sensitized by the Rh positive blood may give birth to abnormal children, if she is married to an Rh positive man. Her children may inherit the Rh antigen from their father. If so the antibodies of the mother may pass through the placenta and cause damage to the red cell of the child during the last few months of pregnancy. This causes the disease known as Erythroblastosis foetalis which consists of an anaemia due to the haemolysis of the red blood cells in the foetus and a consequent jaundice as the blood vessels in the liver become clogged with the broken cells and bile is absorbed by the blood. The blood of the child contains immature nucleated red blood cells (erythroblasts) which are normally found only in the bone-marrow. These cells are not efficient carriers of oxygen. The disease may be so severe as to cause death before birth (stillbirth) or neonatal death (death within a few days after birth). The disease is also known as haemolytic disease because of the haemolysis of the RBCs.

An Rh negative woman may also become sensitized simply by carrying a positive child within her body. Some of the cells from the embryo may seep into her own blood stream through the placental barrier and can cause

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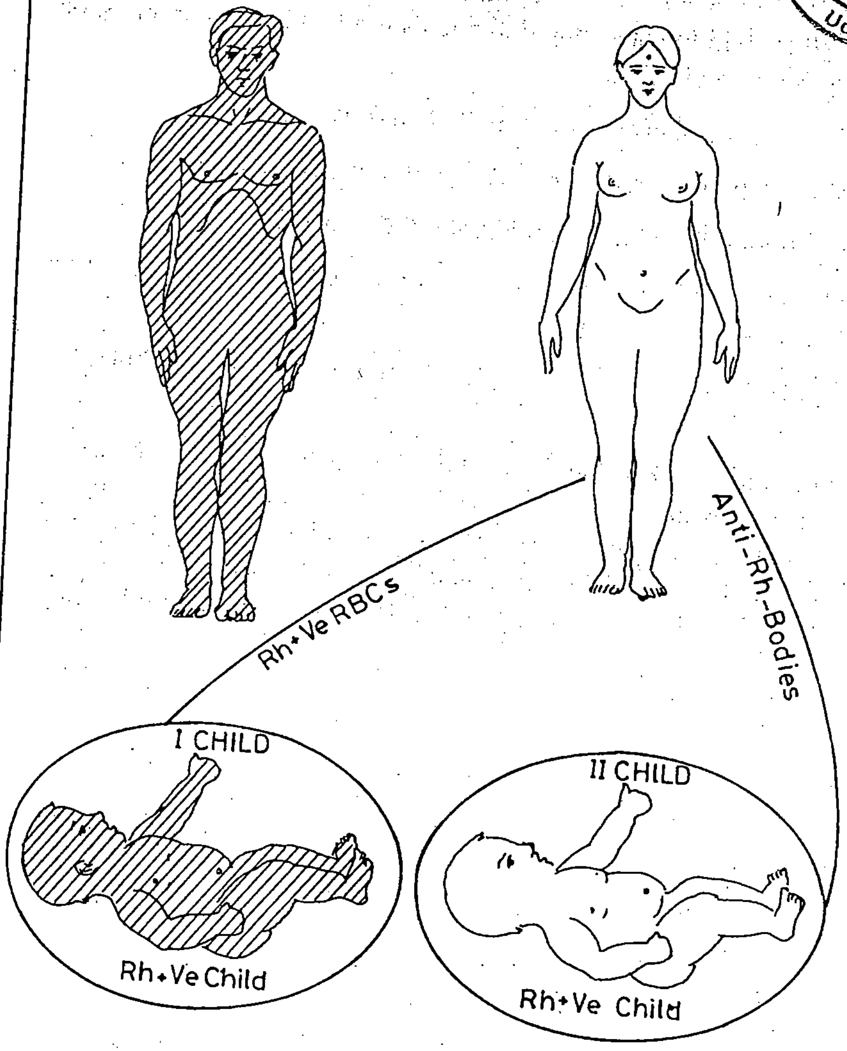
Erythroblastosis Foetalis



II child dies - due to erythroblastosis foetalis.

antibodies to develop. The antibodies do not reach sufficient before the birth of this child to cause any damage as a rule, but subsequent positive children may be affected.

Other classical examples of multiple alleles.

1) Mallard pattern in ducks - Restricted, normal, dusky

2) Feathering in poultry - tandy, retarded, normal

3) Horned condition in certain breeds of sheep horned, knob, small scurs, hornless

4) Production of transferring protein in blood serum lactoglobulin in milk of cattle.

Summing up this chapter, we find that allelic genes are not restricted to occurring in pairs, but may exist in sets of three, four or more. An individual may carry any two of such set of multiple alleles, and a gamete may carry only one at a time involving any two characters dependent upon a set of alleles will always give a one-pair ratio or monohybrid ratio.

EXERCISE

1. Define multiple alleles and write in detail on the inheritance of multiple alleles in rabbits.
2. Discuss on the inheritance of ABO Blood groups in human beings.
3. Write short answers on
   1) Eye colour inheritance in Drosophila
   2) Rh factor
   3) Erythroblastosis foetalis
   4) MN blood groups
   5) Secretor trait
4. A rabbit with a chinchilla coat is crossed with a rabbit with a fully coloured coat. Would it be possible for both albino and Himalayen coats to appear in the offspring? Could either appear?
5. What would be the phonotype of each parent and what kinds of offspring would be produced in each of the following rabbit crosses?

   a) CC X Cc b) $C^{ch}c$ X $C^{ch}c$ c) Cc^{h} X Cc^{h}

   d) $c^{h}c$ X cc e) $c^{h}c$ x $c^{h}c$
6. Suppose a child is of blood group A and the mother is of group O. What group or groups may father belong to?
7. Suppose a father of group A and a mother of group B have a child of group O. What groups are possible in their subsequent children?
8. A woman has a child with erythroblastosis foetalis at her second delivery. She has never had a blood transfusion. On the basis of this information classify the woman, her husband and both children as to Rh blood types.

Chapter 12

Inheritance of Multiple Genes

In the previous chapters, we have studied and discussed the mode of inheritance of characters such as coat colour, eye colour, feather pattern, presence or absence of horn, tall and dwarf plants etc. These traits are sharply divisible, discontinuous in nature and are influenced by one or few pairs of genes. These genes may influence the traits in an independent manner or by dominance, epistasis or by interaction between them.

In contrast, some traits show continuous variation with no dominance between alleles but act in a cumulative manner and have more or less equal effect. Such traits are known as Quantitative traits.

A herd of short horn cows can be easily identified by their coat colour as red, roan and white. But it will be difficult to classify them based on their milk yield. The milk yield grades from a low to a high level.

A group of poultry could be easily identified by the nature of the comb as Walnut, Pea, Rose and Single. But it will be difficult to grade them as high or low egg yielders.

The inheritance of characters such as coat colour in short horns and comb nature in poultry are called Qualitative Inheritance. The inheritance of characters such as milk yield and egg yield are called Quantitative Inheritance. The major differences between these two traits are listed below:

INHERITANCE

	Qualitative Traits	Quantitative Traits
1.	Grade in qualities	Grade in quantities
2.	Discontinuous	Continuous
3.	Depends on one set or a few sets of alleles	Depends upon many sets of alleles
4.	The alleles act in a simple manner of dominance	The alleles act cumulatively. No dominance and the contribution is equal
5.	Sharply divided into two or more clear cut classes	No clear cut classes
6.	Apparent segregation with clear ratios	No apparent segretation and no clear ratios

When several sets of alleles produce more or less equal and cumulative effects on the same character, they are known as Multiple Genes or Multiple Factors or Polygenes.

As early as 1760, Kolreuter, a German Botanist studied hybridization experiments in tabocco plant. Mendel's laws were unknown at that time. Kolreuter did not discover any laws for himself because the characters were quantitative, continuous and not sharply defined. But Kolreuter discovered the important fact that the male and female gametes contribute equally to the hereditary make up of the offspring by conducting reciprocal crosses.

Kolreuter's findings are the same as what we find in the inheritance of quantitative characters i.e. when two parents who differ in a character are crossed, the F_1 are all more or less intermediate between the parents, and the F_2 show many gradations from one extreme to the other. There is no apparent segregation, no sharply defined classes in the F_2 and hence no clear 'ratio'. Kolreuter, then, was completely unable to discover any laws for the inheritance of these character.

The genetic nature of quantitative characters were understood only after simple Mendelian inheritance was discovered 150 years after Kolreuter's pioneer experiment, the same experiment was repeated by East, in tobacco plants.

Among the first crosses of this type which gave a clue to their true nature were those made by Nilsson-Ehle (1908) in wheat. In a cross between two varieties of wheat, one having very dark red kernels and the other white kernels, the F_1 were of an intermediate shades. In the F_2, the colour of the grain ranged from very dark red through various intermediate shades, to white. One, out of every sixteen F_2 kernels was white as the white parent. This immediately reminds us of a modified two-pair ratio, where the double dominant and double recessive each occurs one out of every sixteen individuals on the average. Nilsson-Ehle proposed the hypothesis that two pairs of genes were concerned here, each pair containing one allele which produced some colour and one which produced no colour.

East on the basis of his work on tobacco and corn also suggested a similar hypothesis.

The hypothesis of multiple genes:

Multiple genes form a type of heredity in which no clear cut classes of offsprings are produced. No obvious ratios are found. The F_1 are not like either parent, but are as a rule intermediate between them and are fairly uniform. The F_2 are of all grades, and if enough of them are produced, they range continuously from the extreme represented by one of the original parents to the extreme represented by the other. In the F_2, the extremes are less abundant than the intermediate types.

On this hypothesis of multiple genes, the alleles producing the effect are assumed to be *cumulative* in their action, i.e. two would produce more effect than ones, three would produce more effect than two and so on.

The colour in wheat in Nilsson-Ehle's experiment could be explained as below. Two pairs of alleles A, a and B, b act in such a way, the genes A and B represent the colour producing genes, acting cumulatively. The genes a and b result in no colour production.

Parents	Very dark red AABB	x	White aabb
Gamete	AB		ab
F_1	Medium red AaBb		
F1 x F1	AaBb	x	AaBb
Gamete	AB Ab aB ab		AB Ab aB ab

F_2	AB	Ab	aB	ab
AB	AABB Very dark red	AABb Dark red	AaBB Dark red	AaBb Medium red
Ab	AABb Dark red	AAbb Medium red	AaBb Medium red	Aabb Light red
aB	AaBB Dark red	AaBb Medium red	aaBB Medium red	aaBb Light red
ab	AaBb Medium red	Aabb Light red	aaBb Light red	aabb White

The genotype and their corresponding phenotypes may be listed as below:

Genotype	Numbers	Phenotype	Numbers
AABB	1	Very dark red	1
AABb	2	Dark red	4
AaBB	2		
AaBb		4	
AAbb	1	Medium red	6
aaBB		1	
Aabb	2		
aaBb	2	Light red	4
aabb	1	White	1

Very dark red = 1, Dark red = 4, Medium red = 6, Light red = 4 and White = 1. The F_2 phenotypic ratio is 1:4:6:4:1.

The multiple gene hypothesis in its original form assumes that when two parents produce an intermediate and uniform F_1 and a variable F_2, the genes concerned are equal and cumulative in their effects. The number of pairs of genes concerned is arrived at by an examination of the F_2.

The number of pairs of genes influencing the quantitative traits can be predicted based on the analysis of the F_2 progeny, especially on the proportion of the phenotype of the grand parent which is given in the following table:

Proportion of F_2 extreme as either grand parent	No. of pairs of genes influencing
1/4	1
1/16	2
1/64	3
1/256	4
1/1024	5
1/4n	n

Nilson-Ehle's Experiment in Wheat Multiple Genes Inheritance

Parents	Very dark red X AABB	White aabb
Gamete	AB	ab
F_1	Medium red Aabb	

F2	AB	Ab	aB	ab
AB	AABB Very dark red	AABb Dark red	AaBB Dark red	AaBb Medium red
Ab	AABb Dark red	AAbb Medium red	AaBb Medium red	Aabb Light red
aB	AaBB Dark red	AaBb Medium red	aaBB Medium red	aaBb Light red
ab	AaBb Medium red	Aabb Light red	aaBb Light red	aabb White

Very Dark Red :	Dark Red :	MEdium Red :	Light Red :	White
1	4	6	4	1

If one out of every sixteen is as extreme as each of the original parents, two pairs of genes are concerned, when only one out of sixty four is as extreme as each of the original parents, three pairs of genes must be postulated. The more pairs of genes concerned the smaller will be the proportion of the F_2 like parents.

Another example of multiple factor inheritance could be given in rabbits. The Large Flemish Giant race averages nearly 13 lbs in weight. The small Polish breed averages only about 3 lbs. Crosses between these two races made by Castle produced an intermediate F_1 averaging 7-8 lbs. The F_2 were quite variable, ranging from individuals nearly as small as the Polish breed to those approaching the Flemish Giant in size, but no individuals reaching the extremes of the parental races were obtained in a series of several hundred F_2 individuals. Evidently several pairs of genes are concerned here. It may be assumed that if enough F_2 individuals were raised the parental extremes would eventually be obtained.

	Flemish Giant x 13 lbs	Polish 3 lbs
F_1	7-8 lbs	
F_2	12 lbs	4 lbs

Transgressive variation :

Sometimes, it is found that not only do the parental extremes appear in the F_2, but individuals more extreme than either parent occur. This effect is known as Transgressive variation. In this case, it is necessary to assume that each parent contributed some of the effective alleles. For example in a cross between Seabright Bantams and Golden Hamburgh fowls, which differ considerably in size, Punnet found that the F_1 were intermediate, but the F_2 were so variable as to exhibit extremes beyond the parental weights. While most of the F_2 were more or less intermediate as would be expected, a few F_2 birds were lighter than the Bantams or heavier than the Hamburghs.

Golden Hamburgh	x	Seabright Bantam
1350 gms	750 gms	
1000 gms	650 gms	

F_1 1200 gms

1000 gms

1400 gms	1350 gms	1300 gms	700 gms	650 gms	500 gms

This phenomenon is explained by Punnet on the basis of a four-pair difference between the two parents, the Hamburghs being of the genotype AABBCCdd, while the Bantams may be considered as aabbccDD. The cross is represented as follows :

Golden Hamburgh x Seabright Bantam
AABBCCdd aabbccDD

F_1 AaBbCcDd

AABBCCdd aabbccDD

D contribute more weight. D is called the effective allelle.

Multiple genes in human beings :

In man, the traits height, weight, intelligence and skin colour are governed by the multiple factor inheritances. The differences in skin colour between whites and Negroes appear to be dependent upon multiple genes. Davenport considers that two pairs of genes are responsible for the various grades of colour between black skin and white. The children of Negro and white parents are medium brown in colour and are called 'Mulatto'. When the two parents are Mulattos their children will be of the following colour from the parental extreme to the other namely black, dark brown, medium brown, light brown and white.

Multiple genes in animals :

Milk production in cattle has the multiple gene complex as the basis and it is estimated from 4 or 5 pairs upto 20 pairs. In breeds, as Gurnseys and Holsteins, 10 pairs of genes were found to be responsible for the differences.

Studies on butter fat content in Jerseys and Red Danish cattle has revealed not less than 7 pairs are concened.

Back fat thickness is governed by many pairs of genes and the action is additive and also non-additive.

The egg production in poultry is a complex character and depends upon early and late maturity, egg laying, broodiness, and winter-pause. Distinct multiple genes have been suggested for each form of character and the total of them will make the capacity of egg production.

Modifying genes :

Modifying genes are multiple genes which affect a character which is itself dependent for its initial occurrence upon another gene. For example, the spotted coat such as is found in certain breeds of cattle is due to a recessive gene. Letting S represent the gene for solid colour and s, the gene for spotting, SS and Ss are solid coloured and ss animals are spotted. However, the spotted individuals vary considerably, ranging from those which are nearly solid coloured with a few white patches to those which have only a trace of colour on a white background. Apparently, there are multiple genes influencing the degree of spotting. Since spotting itself is dependent upon a single gene (ss), the multiple genes which affect spotting are called Modifying genes. The gene for solid colour (S) is epistatic over the group of genes which regulate the degree of spotting.

EXERCISE

1. Differentiate between Qualitative and Quantitative traits.
2. Define multiple genes.
3. Discuss on the inheritance of Quantitative characters.
4. Give an account on the hypothesis of multiple genes.
5. Write short answers on

   a) Transgressive variation or

   Effective allele

   b) Modifying genes
6. The mean internode length of a barley variety was found to be 3.20 mm. The mean length in another variety was 2.10 mm. Crossing these two varieties produced an F_1 with average internode length of 2.65 mm. About 6% of F_2 had an internode length of 3.2 mm and another 6% had a length of 2.1 mm. determine the probable number of gene pairs involved in this internode length?
7. A large breed of chicken, the Golden Hamburgh was crossed to small Sebright Bantams. The F_1 was intermediate in size. The mean size of the F_2 was about the same as that of the F_1 but, few individuals were found to exceed the size of either parental type. Explain how can these results be obtained?
8. Who are Mulattoes? What will be the skin colour of the children of two Mulatto parents?
9. What are the genotypes of the parents in the following matings of Negroes?

   a) Medium x light giving one eighth dark and one eigth white.

   b) Medium x light giving one half medium and one half light.
10. Can two Mulattoes have white skinned offspring? Can two white skinned people have dark skinned offspring?

Cytogenetics

कृपया रेषा मारुन
पुस्तक खराब करु नये.

Chapter 18

Chromosomal Aberrations

We know that normally chromosomes occur in pairs in somatic cells and that one of each pair occurs in a gamete. In the course of genetic research numerous individuals, animals and plants have been studied and occasionally variations have been observed in the normal number, structure and arrangement of chromosomes. Such chromosomal aberrations disturb the usual Mendelian ratios and cause the abnormality of the individual.

Classification of chromosomal aberrations:

Two main types of chromosomal aberrations occur and they are as follows:

1. Variations in the normal structure and arrangement of chromosomes. This includes—
    - a. Non-disjunction
    - b. Translocation
    - c. Deletion
    - d. Inversion
    - e. Duplication
2. Variations in the normal number of chromosomes known as Ploidy.

Structural chromosome changes occur as a result of chromosome breakage and reunion of the broken ends in new ways. If both chromatids i.e. whole chromosomes are affected it is a chromosomal aberration if only one chromatid is affected it is a chromatid aberration. When there is no apparent loss or gain of genetic material, the arrangement is considered to be balanced. The inversions, shifts, reciprocal translocation, insertions, etc. are examples of balanced rearrangements.

Non-Disjunction:

One of the first type aberrant behaviour of chromosomes bearing on hereditary factors was worked out by Calvin Bridges on Drosophila. We know that a pair of sex linked factors W and w occur and act in such a way that WW or Ww produce red eyed females while ww produce white eyed females and WY and wY produce red eyed males and white eyed males respectively. When a white eyed female is mated to a red eyed male results are red eyed daughters and white eyed sons.

ww x WY
Ww wy
(red eyed female) (white eyed male)

While this is the usual result expected, Bridges occasionally found unusual offspring resulting from such a cross. For instance, a certain white eyed female when mated to a red eyed male produced not only red eyed daughters and white eyed sons but also a small proportion of White eyed daughters and Red eyed sons.

By reasoning how this might have occurred Bridges finally formulated the suggestion that perhaps XX chromosomes of the female had in some cases not segregated at reduction division. This would result in some of the egg receiving XX chromosomes while an equal number receiving none. Such a female, then in addition to producing w eggs would have produced some with certain factors ww and some with certain no factors of the pair (-).

The results of the different possible fertilizations are indicated in the table:

White eyed x Red eyed
female male
ww WY

Gametes in males W & Y

Suspected gametes w, ww, (-)
in females

	W	Y
w	ww Red eyed female	wY White eyed male
ww	Www Dies	wwY White eyed female
(–)	W- Red eyed male (Sterile)	-Y Dies

Bridges also found that the white eyed females did carry 2 X chromosomes and a Y chromosome and the red eyed males had only an X chromosome with no Y. Red eyed males proved to be sterile. No flies with 3 X chromosomes or with more chromosomes were found indicating that this combination can not live. Bridges named this condition in which both the chromosomes of a pair go to the same cell at reduction division as non-disjunction. It will be noted from the diagram that the white eyed females are females although a Y chromosome is present. Obvisously Y is inert in the female but it is not completely inert in the male because the red eyed males which are males even though Y chromosome is absent are sterile. It is most probably that Y chromosome affects male fertility.

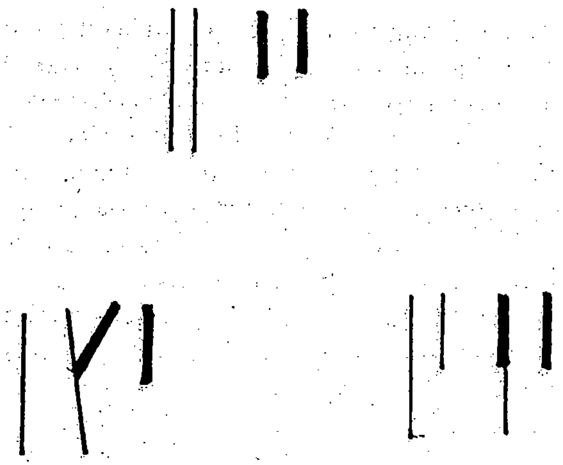
This study of Bridges not only proved non-disjunction but also confirmed that sex linked factors were actually carried on sex chromosomes. In order to further confirm that the factor for white eyes followed the distribution of sex chromosomes, we may mate one of the exceptional white eyed females with a red eyed male and follow the characters and chromosomes of the offspring.

		wwY Exception white eyed female	x	WY Red eyed male
		W		Y
Happened in only 80% of the cases	ww	Www Super ♀ dies		wwY White ♀ eyed
	Y	WY Red eyed ♂		YY Dies
Happened in 92% of the cases	w	Ww Red eyed ♀		wY White eyed ♂
	wY	WwY Red eyed ♀		wYY White eyed ♂

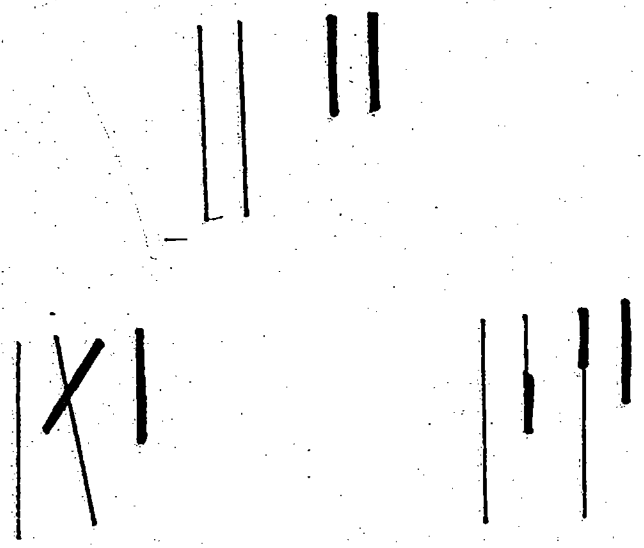
Since the female of the exceptional type is of the genotype wwY segregation of chromosomes at reduction division might result sometimes in eggs of these two constitution i.e. ww and Y and sometimes in eggs of constitution w, wY. Actually, the first two classes were found only in 80% of cases i.e. 2 X chromosomes undergo synopsis more often than do an X and Y chromosome. The results obtained are exactly as predicted in the diagram.

Again red eyed sons and white eyed daughters are produced as exceptions but this time red eyed females contain Y chromosome. Cytological examination reveal that they do. Strangely red eyed males received the Y chromosomes from mother and X from the father which behaviour is opposite to the usual. 1/2 of red eyed females should contain

Translocation



Simple Translocation



Reciprocal Translocation

a Y chromosome and 1/2 of white eyed males should have w Y chromosomes. Cytological examination prove this to be the case.

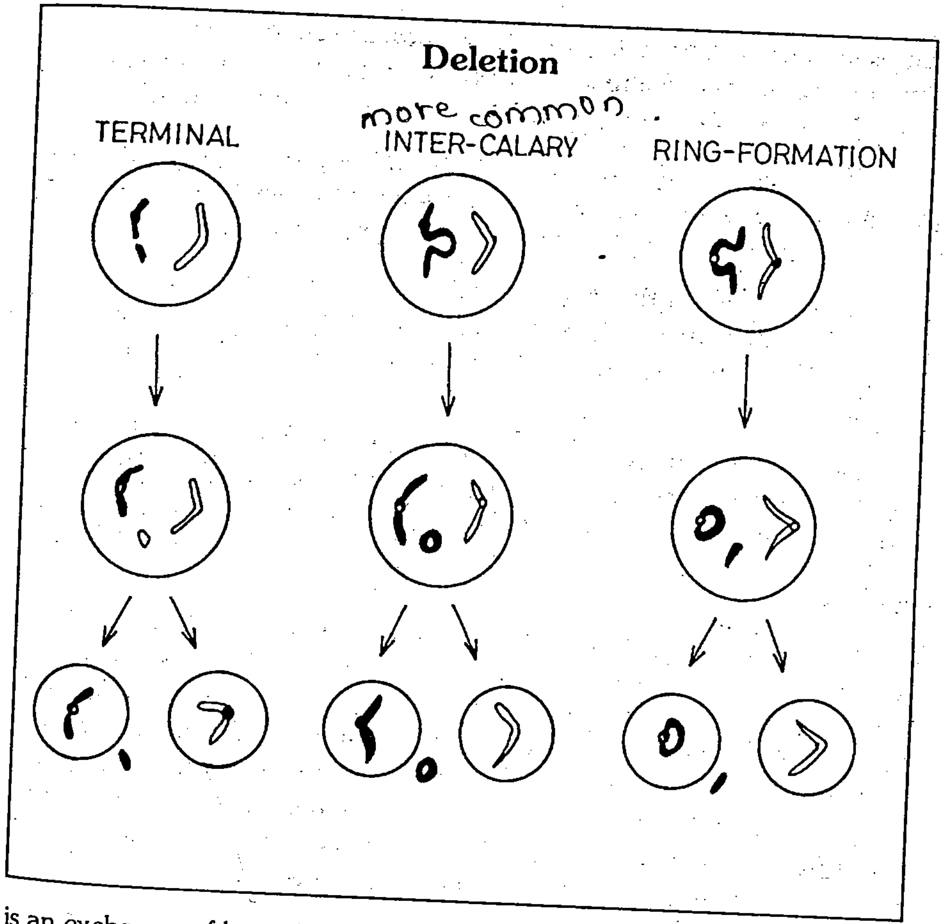
Many cases of non-disjunction are known both in other chromosomes of Drosophila and in chromosomes of other animals and plants. The aberrant behaviour of chromosomes always correlated with corresponding peculiarities in behaviour of the factors. In Jimson weed, Blakesle has noticed many chromosomal aberrations including non-disjunction. This weed has 12 pairs of chromosomes. In addition to normal type, 12 types are recognised, each having one extra chromosome of 1 pair. Thus the chromosome complement of such plant would be 2n + 1 (24+1). Dartington has noticed varieties of cherries which differ from one another in having an extra chromosome of one or another pair. This sort of condition may give rise to important economic variation which in the case of cherries may be perpetuated by grafts.

Translocation:

A piece of chromosome becomes broken and gets attached to another chromosome often of another pair. This process is known as Translocation. ~~They produce~~ detectable genetic abnormality and can also be confirmed cytologically.

For instance, in Drosophila, a few genes in the third linkage group were found by Muller to be linked to the genes on the second linkage group which was an aberrant condition. The genetic evidence indicated that a large block of genes from the third linkage group had somehow become a part of the second linkage group. Therefore, if the genes are really carried on chromosomes, the cytological examinations of chromosomes should show a translocation of part of a chromosome. Cytological examination revealed one of the third chromosomes to be shorter. Correspondingly one of the second chromosomes was longer than usual. The aberrant behaviour of the factors was directly correlated with an aberration in the chromosomal structure.

Reciprocal translocations are more frequently met with than the Simple Translocations. ~~When there~~ is mutual exchange of parts by non-homologous chromosomes, it is known as Reciprocal Translocation. Crossing over also involves an exchange of material between two chromosomes, but here, there is an exchange of homologous parts between homologous chromosomes. In reciprocal translocation, the exchange of parts is often between the chromosomes belonging to different pairs. The results are of considerable importance in the production of new varieties.



Various theories have been put forth for the occurrence of Reciprocal Translocation. Some attribute this to fragmentation of chromosomes during gametogenesis which gets attached to produce new combinations. The second view is that translocation occurs due to overlapping, entanglement and subsequent abnormal breaking with non-homologous exchange of parts.

Due to reciprocal translocation, in synapsis, the pairing could not be complete, since only homologous regions undergo synopsis. Hence, many abnormal synoptic configurations like rings and chains of chromosomes are seen. The formation of rings of chromosomes is called 'Catenation'.

Like alleles attract each other while the unlike alleles repel each other. Hence, homologous bits of chromosomes seek union and this explains the cause of formation of rings and various configuration in the cells.

Centric fusions or Robertsonian translocations occur between the two acrocentric chromosomes when breaks occur close to the centromere in the long arm of one and short arm of the other, followed by exchange and reunion.

Tandem fusion type of translocations occurs when there are breaks close to the centromere in one chromosome and close to the end of arm in another followed by an exchange and reunion.

Deletion:

It is common for a portion of a chromosome to break. When a broken piece is devoid of the centromere, it does not move towards the pole along with the spindle fibres and gets lost in the cytoplasm. The genes carried in the broken bit is also lost. This phenomenon is known as Deletion. Deletion does not affect the viability of an individual, if the missing piece is not large. When only a small portion is deleted the individual lacking it may survive especially in the heterozygous conditions. With a similar deletion on both the members of a pair, the individual will rarely live.

An interesting example of deletion in which the parallel cytological and genetic facts have been investigated, is that of the loss of a gene in mice. In these animals, the gene for normal gait V is dominant to the gene for waltzing gait v. Waltzing mice are unable to run in a straight line and usually run about in small circles. Crosses between homozygous normal mice VV and homozygous waltzers vv result in normal offspring Vv. In one such cross, however, a waltzing female was produced, which was observed by Gates in his laboratory.

He concluded that the gene for normal gait had been lost from this individual either as the loss of the entire chromosome carrying this gene by non-disjunction or as a loss of a part of the chromosomes carrying the gene

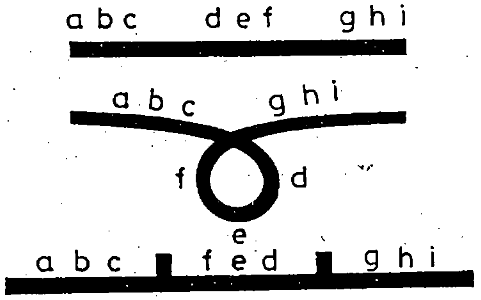
for normal gait (V) by deletion. When the chromosomes were examined, it was found that in this mouse and others like it produced by suitable breeding, it was also found that one member of a pair of autosomes was only of quarter size in length as compared to its mate. In normal mice, the members of this pair of chromosomes were found to be equal in size. The evidence from deletion proves that genes are actually carried on the chromosomes.

Two types of deletions are possible. A terminal deletion in which a portion is lost from the end of the chromosome. An Intercalary deletion in which a portion of the chromosome is lost from within leaving the ends of the chromosomes intact. This type of deletion may occur when a chromosome breaks in two places and the end pieces become attached to each other. The two ends of the portions deleted from the centre of the chromosome may become attached to form a ring shape structure. Such ring chromosomes are unstable and can not be maintained, because of abnormal meiosis. The Intercalary Deletions are by far the more common of the two types.

Inversion:

Occasionally, a portion of the chromosome will break to produce a deletion but the broken portion will become re-attached to its original portion with the two ends reversed. This is most likely to happen, the chromosome

Inversion



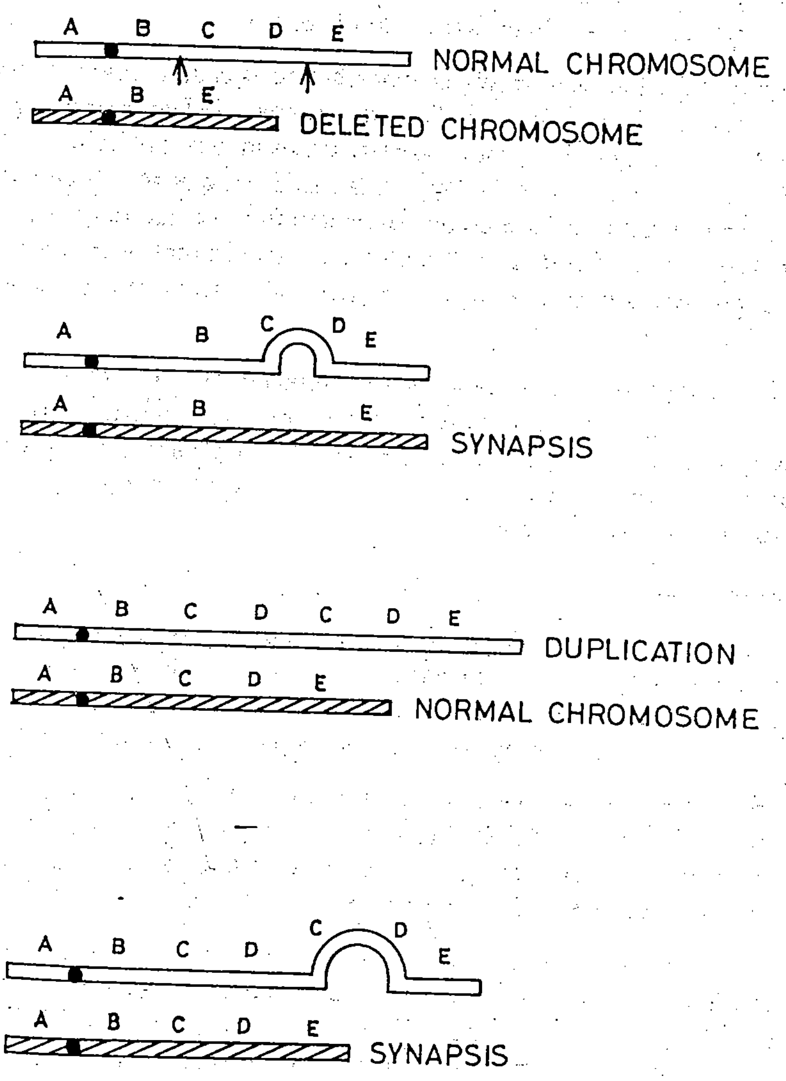
The sequence of genes reversed due to change in the position of a portion of chromosome forms something of a loop since such twisting makes easy for the broken ends of the deleted portion to become reattached in the new position. Inversion is most likely to occur in meiosis and will result in germ cell carrying chromosome with genes in reverse order on the inverted portion.

Let us suppose that the original chromosome carried genes a b c d e f g h i. In that particular sequence and the inversion of the central portion of the chromosome occurred, the new sequence might be a b c, f e d, g h i. The location of the three central genes would no longer conform to their position as previously determined by chromosome mapping. When a germ cell carrying such a chromosome unites with normal germ cell, there is usually no detectable effect of the inversion on the phenotype of the offspring for all the genes are present in normal number. When synapsis in subsequent generations, however, the cytological figures show abnormal configuration. During the gametogenesis, perfect synapsis is not possible and crossing over can not take place. The giant chromosome of Drosophila produce a remarkable way for quickly recognizing inversions. Quite often, a geneticist may wish to maintain a series of genes on a chromosome in a heterozygous state without any crossing over. This is possible, provided there is an inversion on one of the two paired chromosomes. Such an inverted X chromosome known as C1B is very valuable in mutation studies.

Probably, inversions also play a part in evolution of separate species, just as translocations do. Inversion also proves that the genes are arranged in a linear order on the chromosomes.

Duplication:

Large or small pieces of chromosomes containing extra blocks of genes due to duplication are found in many individuals or even in races. It is assumed to arise as a result of unequal crossing over of the synaptic chromosomes of primary gametocytes. In such a way, that one chromosome suffers a deletion and the other receives a duplication of the short segment. This method furnishes an obvious means of accumulating additional chromosomal material in an organism. Having once become homozygous a duplicated portion can be transferred to other position within the same chromosome or to a different chromosome as a result of inversion or



Synapsis of Normal and Deleted and Duplicated Chromosome

translocation. According to Painter, duplications are common and are of great significance in evolutionary origin of species.

One of the best known duplications in Drosophila, is that resulting in the bar eye condition. Bar eyed flies have narrow slit like eyes in place of normal oval eyes and abnormality is inherited as though due to a sex linked gene without dominance, heterozygous female having eyes of intermediate width. Sturtevant and Morgan demonstrated that the bar gene is really a short duplication. Bridges confirmed this by cytological examinations of the giant chromosomes showing that the duplicated segment is in the region of X chromosome.

Homozygous bar eyed females possess this segment twice in each sex chromosome whereas normal females have it singly in each. By usual crossing over in bar eye female it is possible to obtain this segment in triplicate, a condition which results in an even more extreme reduction in eye size known as Ultra bar.

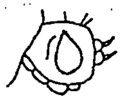
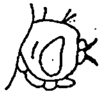
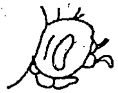
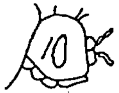
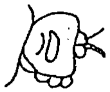
Position Effect:

The effect of a gene depends not only upon its presence but also upon its position with respect to its neighbouring genes. This is known as Position Effect. If, for example, we designate the duplication responsible for bar eye as B the genotype of the various individuals may be written as follows:

BY, Normal male. BBY, bar eyed male. BBBY, ultra bar eye male. BB, normal female. BB B, heterozygous bar eyed female. BB BB, homozygous bar eyed female. BBB B or BBB BB heterozygous ultra bar eyed female and BBB BBB, homozygous ultra bar eyed female.

In general, eye size decreases with the number of duplicated segments. The position of these segments on the chromosomes however, is extremely important in producing this effect. Thus, a male having two bar eye segment on one chromosome (BBY) has much smaller eyes than a female having one bar eye segment on each of the two chromosomes (BB). The eyes of an ultra bar male (BBBY) are much more reduced than those of a heterozygous bar eyed female (BB B). Within the same sex, the eyes of heterozygous ultra bar females (BBB B) are smaller than those of homozygous bar eyed females (BB BB) although the number of segments

Position Effect

X Chromosome		Phenotype
B/B	Normal	
BB/B	Heterozygous	
BB/BB	Homozygous Bar Eye	
BBB/B	Heterozygous Ultra Bar	
BBB/BBB	Homozygous Ultrabar	

Genotypes and Phenotypes of Bar Eye in Drosophila

is the same.

Thus, the effects of the genes depend not only upon their number and kind, but also upon their position.

Ploidy:

A general term for the variation in the number of chromosomes is Ploidy. Aneuploidy is applied for those involving addition of one or more chromosomes or pairs, but not the whole compliment. Occasionally, there is an increase in chromosome number, due to one or more complete sets of chromosomes. It is known as Euploidy or Polyploidy. Individuals with 2n + 1 chromosomes are called Trisomics. Those with 2n + more than one i.e. 2n + 1 + 1 are called Polysomics.

Polyploidy:

We know, now, through non-disjunctions one or more times so that an individual may have 3 of each kind of chromosomes or 4, or more. Such individuals are said to be Polyploidy. If there are 3 of each kind of chromosomes, individual is called Triploid. If 4, a Tetraploid and so on. Thus, the haploid number may be duplicated as many times as 12 or 15 times in some varieties. Polyploidy is more common in plants than in animals.

In the vicinity of Scar tissue produced as a result of cutting or grafting, the adventitious shoots, which grow out are Tetraploids. In these cases, this chromosome has apparently split as in mitosis but the cells have not divided resulting in cells with 4n number of chromosomes which thereafter undergo mitosis, in regular fashion and repeat the 4n number. Blakesle has proved that colchicine when applied to seeds or buds causes a large proportion of the cells to become a tetraploid. Colchicine acts as a mitotic inhibiter particularly at prophase stage. Here, again, the polyploid condition arises as a result of splitting of chromosomes as in mitosis with a corresponding failure of the formation of cell wall.

Polyploids arise from a duplication of chromosome complement within the same species and they are known as Autopolyploids in contrast to Allopolyploids which arise from hybridizations of two species or genera with

subsequent duplications of each chromosome complement. The reproduction of these two types varies.

Autotetraploids have four identical chromosomes of each kind. Hence, in synapsis, all four of each sort may come together so that normal segregation does not necessarily take place. Often, two chromosomes of each kind go to each pole. But sometimes, the segregation of three to one pole and one to the other may take place. Thus, some of the gametes of an autotetraploid have chromosomes in excess while others are deficient in certain chromosomes and fertilization involving these peculiar gametes results in less viable or inviable offspring. In this manner, the fertility of autotetraploids may be reduced. Autotetraploids are larger than the corresponding diploids in practically all parts. (Height somehow is not affected) Since each chromosome is duplicated no specific character is affected than the others.

The reproductive situation is different in allotetraploids. Here, the two diploid complements of chromosomes come from different species or different genera. Therefore each chromosome has only one mate. Synapsis and segregations are normal and the new types breed true without any reduction in fertility. Allotetraploids, generally arise through hybridizations from union of a haploid set of chromosomes from each of two taxonomically different individuals (species) with the subsequent doubling of the chromosomes in occasional gametes or the hybrid. Many, if not in most species, hybrids are sterile but in few cases a few diploid gametes are produced and when two such diploid gametes unite allotetraploids arise having characters different in many ways from either parent species. In this way, fertile new species arise. In order for the tetraploid to breed true it must produce diploid gametes. In order for any polyploid variety or species do breed true it must have chromsomes of each kind in even numbers so that regular segregation can occur. Thus triploids and pentaploids and others having each kind of chromosomes in odd numbers can not breed true. In these cases, a large amount of sterility occurs with the production of offspring having one or more chromosomes in excess of the normal numbers.

Triploids may come about through the union of a gamete from a tetraploid with a gamete from a diploid. Eventhough a triploid plant can not breed true, it may be propagated asexually.

Not only have new autopolyploid and allopolyploid species and varieties been experimentally produced but there is evidence that they occur in nature and that they are a potent factor in evolution. Many cases of alloploidy in nature are known particularly among the chestnuts, grasses and roses. Various cultivated varieties of garden flowers, vegetables, crop and fruit plants are all polyploid (Tulips and Lillies). Polyploidy is also of economic importance in that it has yielded varieties of cotton. Triploid and tetraploid tomatoes have been produced.

The expression of the genetic factors varies among polyploids. In the case of certain genes, one dominant factor may express itself in the presence of two or even more recessive alleles. In the case of other factors one dominant may not be able to produce its usual effect in the presence of 2 or even more recessive alleles. Genetic ratios are thus affected in several ways.

It is interesting to observe that related species of genera frequently have chromosome numbers that are multiples of basic number indicating that the species or genera have differentiated in nature through polyploidy. Thus, the common species of wheat has 7 pairs, 14 pairs, and 21 pairs. In Chrysanthemums, the number range from 9 pairs to 45 pairs. Sometimes, related species differ by only 1 or 2 chromosomes indicating that non-disjunction may also be effective in evolution. Compared with the slow accumulation of gene mutations, Ploidy is a sudden process and evolution by this means is often called Cataclysmic Evolution.

Chimeras and mosaics :

A change in chromosome number is not necessarily restricted to zygotic changes which affect all cells of an individual. Occasionally a chromosome change occurs after the zygote has been formed which may lead to sections of tissue growing side by side bearing different chromosome constitutions. Such individuals are known as chimeras (after a mythical monster with the head of lion, the body of goat, and a serpent's tail) or mosaics; and arise either spontaneously or through chemical and physical treatment. In mammals an embryonic transfer of blood forming tissue may occur between fratrnal dizygotic twins leading to the presence of more than one type of blood tissue in an individual. Chimeras are this individual's possession genetically distinct tissue.

Karyotyping:

Metaphase chromosomes can be drawn or photographed and arranged in homologous pairs in a systematic manner to form a karyotype. A diagrammatic representation of chromosome morphology is known as idiogram.

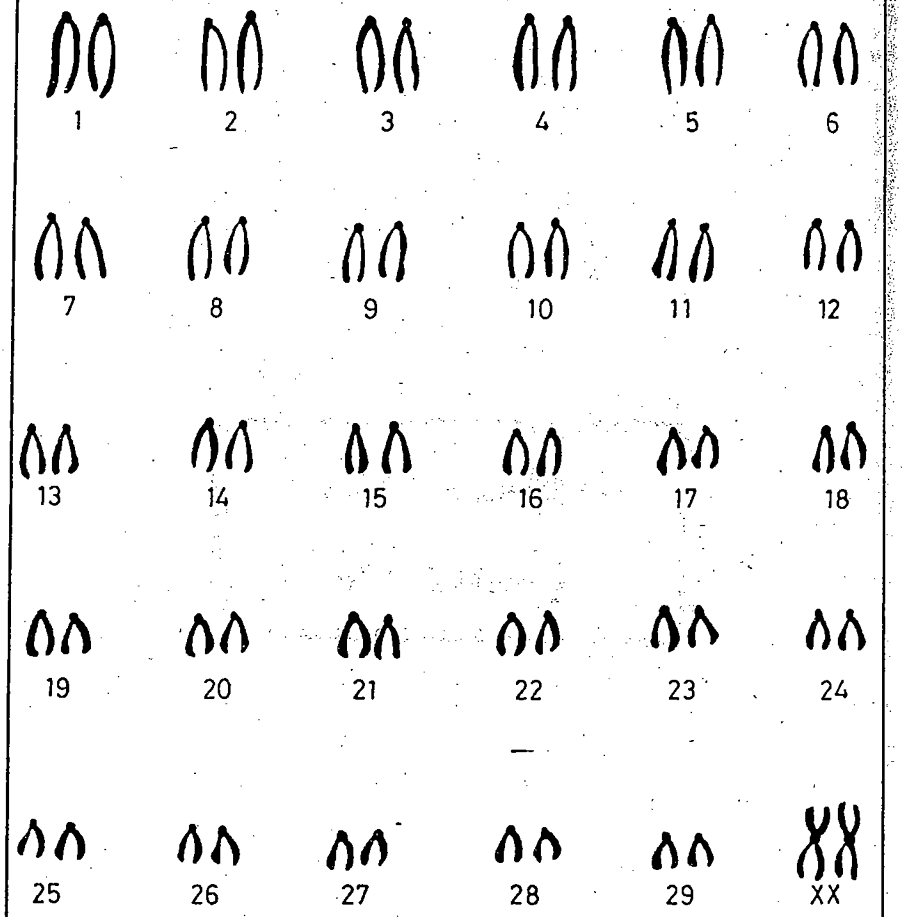
During metaphase stage of mitosis the chromatids become shorter and thicker, and so the number, size and morphology of chromosomes can be observed under light microscope following appropriate treatment of the cells. Their appearance is characteristic for each species. There are two sets of chromosomes in somatic cells constituting the diploid or 2n number.

The diploid number for various domestic species are, horse-64, cattle-60, buffalo-50, sheep-54, goat-60, pig-38, dog-78 and cat-38. The size of the chromosome is dependent on the length of its arms, while its shape is dependent upon the position of the centromere which is seem as a constriction. At metaphase each arm consists of two chromatids lying side by side. When arms are equal in length, or almost so the chromosome is metacentric. When one arm is only one-half to one-third as long as the other, the chromosome is submetacentric. When one arm is only one-third to one-seventh as long as the other chromosome is of subtelocentric. When the centromere is very close to the end of the chromosome, it is acrocentric and when the centromere is at the end, it is telocentric. The short arm is designated as P and the long arm is Q.

Chromosomes in man : In 1956 Tjio and Levan demonstrated conclusively that there are 46 chromosomes in man, or 23 pairs, of which 22 pairs are autosomes and one pair of sex-chromosome, males having X, Y and females two Xs.

Bacterial Genetics

Karyotype of Cow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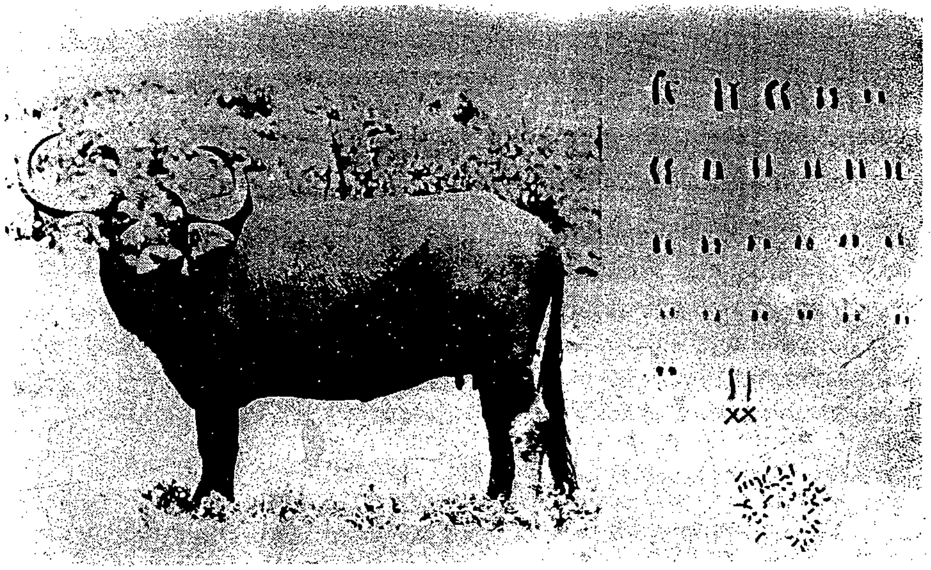
Karyotype of a She Buffalo



Depending on size and centromere in position the human chromosome complement is grouped as in the following table:

Group	Size	Centromere location
1-3	large	approximately median
4-5	large	submedian
6-12 & X	medium	submedian
13-15	medium	acrocentric
16-18	short medium	median or submedian
19-20	short	approximately median
21-22 & Y	very short	acrocentric

Distinction between chromosomes within a group may be quite difficult.

of a single bacterium are genetically identical and they are referred to as a clone or colony visible to the nachecked eye when grown on a solid medium like agar plate.

The disadvantages of this study on microorganisms are dearth of regular recombinational events. Asexual reproduction is by amitosis a direct nuclear division without the formation of condensed chromosomes or spindle fibres. Another disadvantage is the relative lack of morphological variation in micro- organisms.

Genetically different clones may respond differentially to nutrients dyes, drugs or pathogens in a culture medium. Wild type strains are able to grow on minimal medium - prototrophic whereas some mutant strains require supplementation - auxotrophic. Bacteria and viruses do not have easily visible chromosomes like higher organisms. But even the morphology of viruses can be studied now through the electron microscope.

Genetic recombination in bacteria:

Several mechanisms exist in bacteria which can effect genetic recombination.

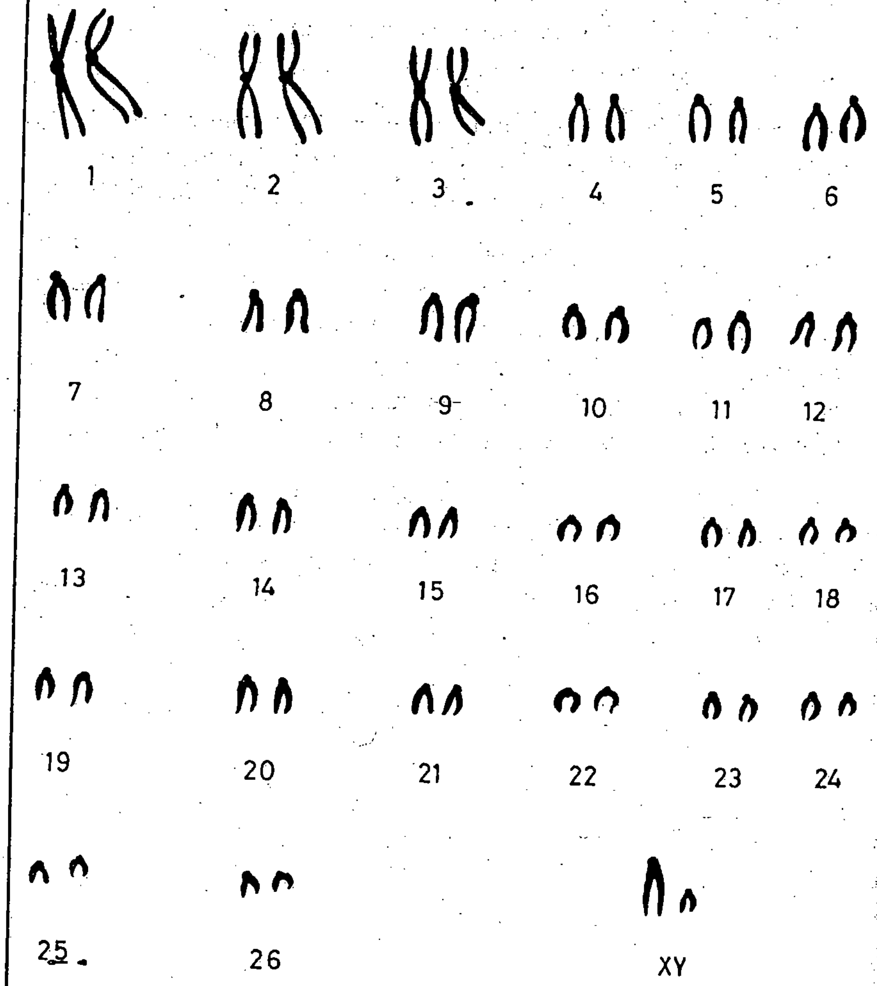
1) *Transformation* - Nacked DNA molecules from one bacterial strain can be absorbed by another strain and utilized in the production of a new recombinant phenotype. For example when pneumococci are surrounded by polysaccharide capsules they are virulent and capable of producing disease and form colonies with a smooth border on soild medium. Unencapsulated pneumococci are avirulent and plate as rough shaped colonies. When a rough strain is exposed to a purified DNA extract from a smooth strain, some rough cells are transformed into smooth cells. This is a evidence for the proof that DNA is genetic material. Transformation may occur in some species with frequencies as high as 25% but generally occurs at low frequencies.

2) *Conjugation* - It is the transfer of genetic information through the formation of a cytoplasmic bridge from one cell to another. Conjugation is a one way transfer of DNA from a donor cell to a recipient cell. In Escherichia coli strain K-12, the male possesses may replicas of the sex factor F which is symbolized F+ when the sex factor is in its extrachromosomal phase. Females do not have the sex factor and are symbolized F-. When F+ cells conjugate with F- cells, one or more sex factors are transmitted across the cytoplasmic bridge to infect the F- recipients and convert them into F+ (male) cells.

3) *Transduction* - It is the transfer of gene from one bacterium to another mediated by a virus. Bacterial viruses are called bacteriophage. Lambda phage in its host E. coli is the best example of bacteriophage.

4) *Sexduction* - The bacterial genes carried from one cell to another on a molecule of DNA which acts as a sex factor called 'F'. This sex gene can reside in a bacterial chromosome or it may exist as an autonomous unit extra chromosomally. Sexduction is also referred as F duction. Genetic elements which possess the dual capacity to exist either as a chromosomal or an extrachromosomal entity are called episomes.

Thus even in lower organisms like bacteria and viruses we find the possibility for the exchange of genetic material and for the formation of new species. Further the study of the genetics of bacteria and viruses is opening up new avenues of the chemical and physiological basis of the units of heredity.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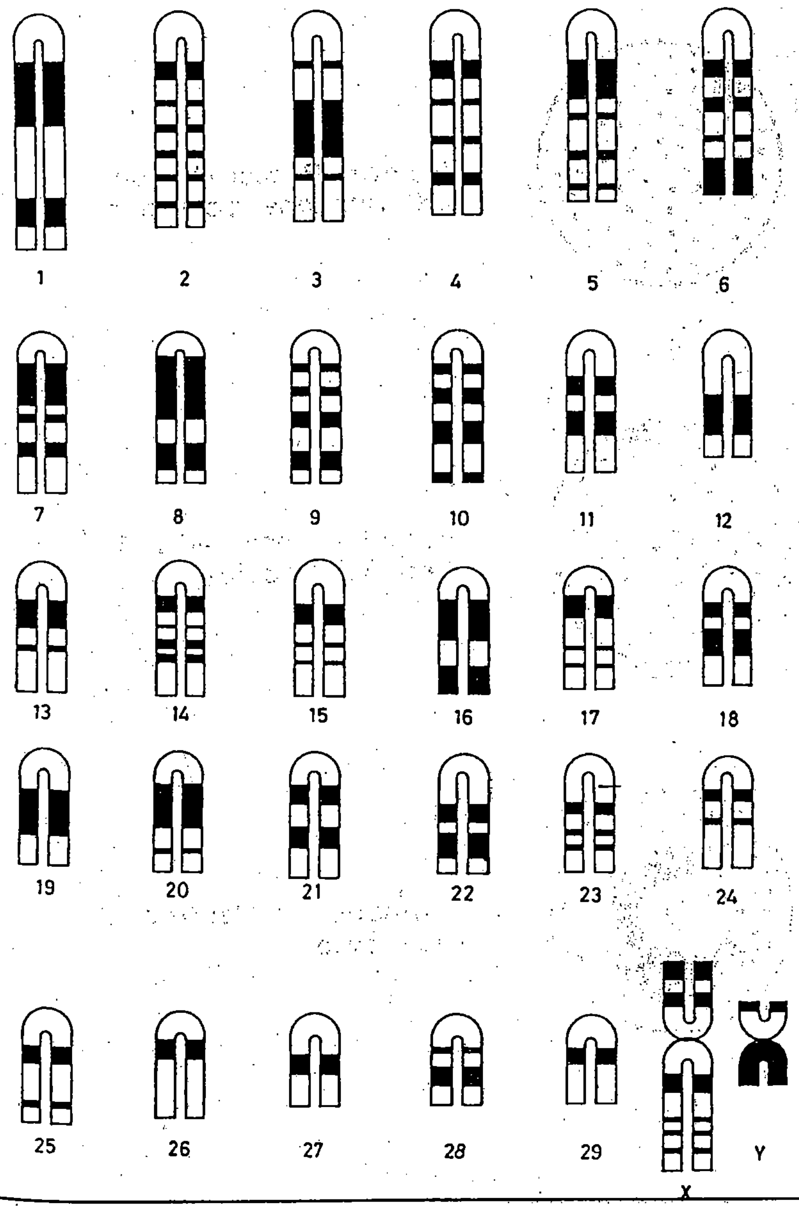
**Karyotype of Ram
-C-banded chromosomes**

Diagram showing G-Bands in Cattle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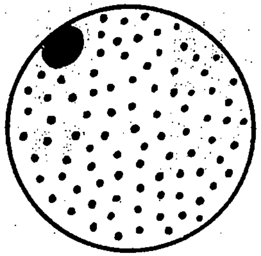
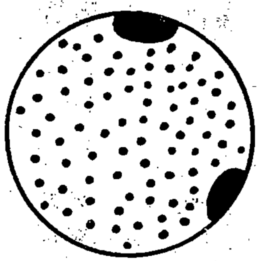
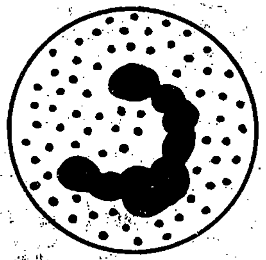
Sex Chromatin



Human Buccal Mucosa cell with one barr body



Buccal Mucosa cell with two barr bodies



Drumstick in Neutrophilic Leukocyte

Normal Chromosomal complement of domestic animals

Sl. No.	Animal	Diploid No. (2N)	Autosomal pairs				Sex chromosome	
			AC	MC	SMC	TC	X	Y
1.	Cattle (B. taurus)	60	29	-	-	-	MC	MC
2.	Cattle (B. indicus)	60	29	-	-	-	MC	AC
3.	River buffalo	50	19	-	5	-	AC	AC
4.	Swamp buffalo	48	18	1	4	-	AC	AC
5.	Horse	64	18	-	13	-	SMC	AC
6.	Pig	38	6	5	5	2	MC	MC
7.	Goat	60	29	-	-	-	AC	MC
8.	Sheep	54	23	3	-	-	AC	AC
9.	Dog	78	38	-	-	-	MC	MC
10.	Cat	38	2	16	-	-	MC	MC

AC - Acrocentric; MC - Metacentric; SMC-Sub-metacentric; TC - Telocentric

Chromosome banding Techniques

A band is defined as a part of chromosome which is clearly distinguishable from its adjacent segments by appearing darker or lighter. There are different types of bands. The bands stained with basic dyes such as Giemsa are called G bands. Those bands of chromatin stained by methods that demonstrate constituve heterochromatin are called C bands which are mainly confined to centromeric region. The bands seen with fluorescent dye like Quinacrine are called Q bands. The development of banding techniques Q, G, R, C and T banding producing staining patterns that are characteristic of each chromosome pair has made it possible to karyotype and identify structural rearrangements and deletions with much more greater accuracy.

Uses of banding techniques :

The banding techniques are used for the various studies such as studies on Y chromosome, identification of trisomics and studies on the cancer cells. Further it is useful for the detailed study of the morphology of the chromosomes and the abberations if any.

Types of banding:

1) *Q banding* : The principle involves in this technique is the staining of cells with flurochromes and exposing them to the U.V. light of a fluorescence microscope. Quinacrine mustard and Quinacrine dihydrochloride compounds are used in this technique. The prime prerequisite for obtaining clear banding patterns is well spread chromosomes with fairly long chromatids held closely together. It necessitates the use of fluorescence microscope, and does not permit permanent preparation. In this technique a characteristic intensive fluorescence is observed in distal half of the long arm of the Y chromosome.

2) *G banding* : G bands are the cross bands of various width and shades stained with Giemsa or similar stains. The G banding technique produce permanent bands and do not require special optical equipment.

3) *C banding* : Constructive heterochromatin is a relatively well defined fraction of chromatin usually located around the centromere or in blocks in the chromosomal arms. C banding techniques selectively stain chromosomal areas known to contain constitutive heterochromatin, mainly of the pericentromeric type. The variation in size of C bands may correspond to different amounts of satellite DNA present.

4) *Reverse and Telomeric bands (R and T)* : This is a technique in which there is reverse of those formed by Q and G banding techniques. Thus G and Q negatively stained bands appear positively stained and vice versa. These R and T techniques have the particular advantage of showing very clearly the staining of telomeric areas of chromosomes which are difficult to observe with the Q and G bands because of their light staining.

5) *Feulgan (F) bands* : The Feulgan stain is an old and well known specific DNA stain. The bands resemble those elicited by Q and G banding techniques. The bands resulting from the use of the Feulgan have been named F bands. Several telomeric segments that Q and G negative, stain positively with the F banding technique. The distal half of

the human Y chromosome which is brightly fluorescent with Q bands and intensively stained with G bands appears negative under F banding technique.

Of all the banding techniques Q, G and R bands have been widely used for the study of chromosomal abnormalities.

The proof of the hypothesis that hereditary factors are carried on the chromosomes:

1) Hereditary factors are carried in the sperms or the eggs or both since only these bridge the gap between generations. Although in some species the embryo develops within the body of the mother during gestation, principles of heredity appear to be same for all species including those which have external fertilization and development.

2) Within a species, the sperm and egg (with the exception as in sex linked inheritance, aberrations) contribute equally to the inheritance of specifications, proved through reciprocal crosses.

3) Although the egg has a relatively large amount of cytoplasm in addition to nucleus, the sperm is practically all nucleus. Only nuclei unite at actual fertilization, and this is the essential part of the gamete in regard to transmission of genes.

4) Of the nuclear constituents, only the chromatin material appears to be accurately divided at mitosis and segregated during maturation.

Parallels between the behaviour of factors as seen in results of breeding and behaviour of chromosomes as seen under microscope are listed below:

1) Factors normally occur in pairs in the cells of the individual so do chromosomes.

2) The two factors of each pair segregate in the formation of the germ cells. The two chromosomes of each pair also segregate in the formation of germ cells.

3) Certain factors assort at random, likewise certain parts of the chromatin material (that is, whole chromosomes) assort at random.

4) Certain factors behave as though only one member of the pair were present in one sex (sex linked factors). Similarly only one member (or at least one normal member) of one pair of chromosomes is present

in the corresponding sex.

5) Certain factors do not assort at random but occur in paired groups (linkage groups) which tend to be transmitted as units. The chromatin material is also gathered into paired groups (chromosomes) which tend to be transmitted as units.

6) The members of linkage group do not stay completely together as a rule, but during maturation exchange with a definite frequency homologous members of the paired groups (genetic crossing over). The pairs of chromosomes also exchange homologous parts of their lengths during maturation of germ cells (cytological crossing over).

7) In certain cases genetic crossing over is more frequent in one sex than in the other. In these cases, chiasmata formation is proportionately more frequent in the sex which exhibits more crossing over.

8) At the time of genetic crossing over, the factors are arranged in a specific linear order. At the time of cytological crossing over, the chromatin material is in an attenuated linear arrangement.

9) The number of linkage groups as a rule definite and constant for any species and do not exceed the number of pairs of chromosome.

10) Factors occasionally behave in a peculiar unexpected ways (abnormal ratios, unusual linkage relationships, genetic deficiency). In these cases the chromosome are also found to be aberrant (non-disjunction, translocation deletion, catenation, ploidy etc.).

It would appear as an unescapable inference from the above facts and principles that hereditary factors are carried in the chromosomes.

EXERCISE

1. What is meant by chromosomal aberration? Classify it.
2. What are the structural chromosome abnormalities?
3. Discuss on
    1) Non-disjunction
    2) Translocation
    3) Deletion
    4) Duplication
    5) Inversion
    6) Position effect
4. What is meant by Ploidy and classify it?
5. Write short answers on
    a) Aneuploidy
    b) Polyploidy
    c) Chimeras
    d) Klinefelters syndrome
    e) Turner's syndrome
    f) Down's syndrome
    g) Karyotyping
    h) Morphology of chromosomes
    i) Banding techniques
    k) Sex chromatin

6. Give the proof of the hypothesis that hereditary factors are carried on the chromosome.
7. Discuss the significance and role of Polyploidy in evolution.
8. The diploid chromosome number of an organism is 12. How many chromosomes would be expected in

   a) a Monosomic b) a trisomic c) a tetrasomic

   d) a nullisomic e) a monoploid f) a triploid

   g) an autotetraploid
9. What is the role of duplication in evolution?
10. A chromosome with segments in the normal order is abcdefgh. An inversion heterozygote has the abnormal order abfedcgh. A three strand double cross over occurs involving the regions between a and b and between d and e. Diagram and label the first and second anaphase figures.

कृपया रेषा मारुन
पुस्तक खराब करु नये.

Chapter 19

Determination of Sex

Sex is the fundamental quality which is recognised in living things, particularly in the higher forms of life.

Importance of sex in Heredity:

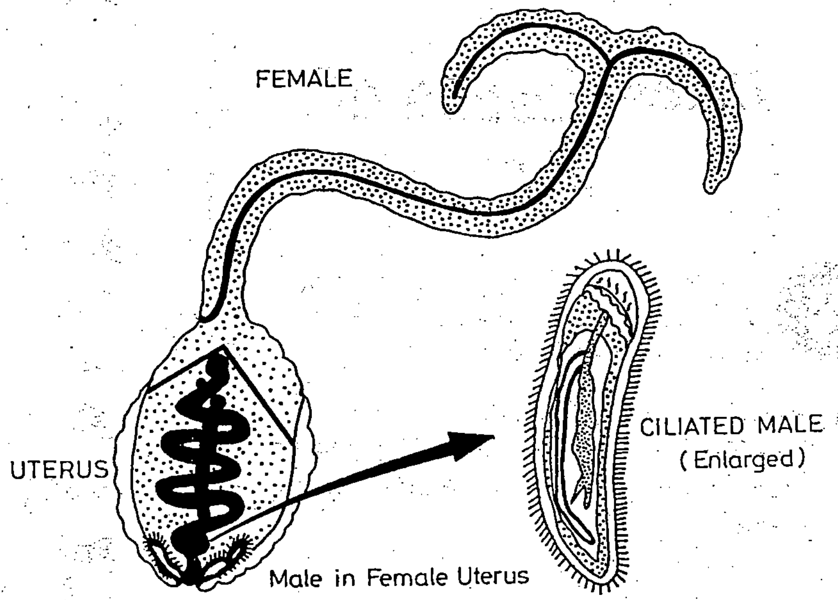
1. Sex is the main source through which genetic variation can be brought about between one generation to the next.
2. Sex is the only way to maintain the germplasm.
3. Sex provides the way to unlock the study of the secrets of chromosome behaviour.
4. Sex serves to promote hybrid vigour.

Mechanism of sex determination:

There are three suppositions by which sex can be determined. They are:

1. ***Progamic:*** i.e The sex of the offspring is determined before fertilization. This support is true in case of the sex determination of birds, moths, butterflies and some fishes. The females are heterogametic and males are ho-

Sex Determination by Environment



Environment - Determines the Sex

Worms in Water - Becomes Female

Worms in Uterus - Becomes Male

mogametic. The sex of the offspring is determined by the nature of chromosome of the ovum i.e. sex is fixed even at gametic formation.

2. ***Syngamic:*** i.e. The sex of the offspring is determined at the time of fertilization. This is true in case of the sex determination mechanism of Drosophila and mammals where the females are homogametic and the males are heterogametic. The sex of the offspring depends upon the nature of the chromosome of the sperm.

3. ***Epigamic:*** i.e. The sex of the offspring is determined after fertilization. The environment can not modify the sex in some higher forms of life. But it can determine the sex in some lower forms of organisms like the marine worm Bonnelia Virdis. In bonnelia sexual dimorphism is seen. The female has a long bifurcated probosis about 3 feet in length and a body of about 2 to 5 inches. The male is ciliated and very small about 1 mm in length and lives in the oviduct of the female. The fertilized eggs become tiny free swimming larvae. The larvae which settles in the mud become a female. If the free swimming larva gets a chance to come into contact with the probosis of the female, it gets attached to the probosis eventually penetrates into the oviduct and becomes a male. Culture studies reveal that it takes 4-5 days for the male to get matured on the probosis of the female. The water environment makes the larva to be female and the environment in the probosis makes the larva a male.

THEORIES OF SEX DETERMINATION

XY chromosomes method:

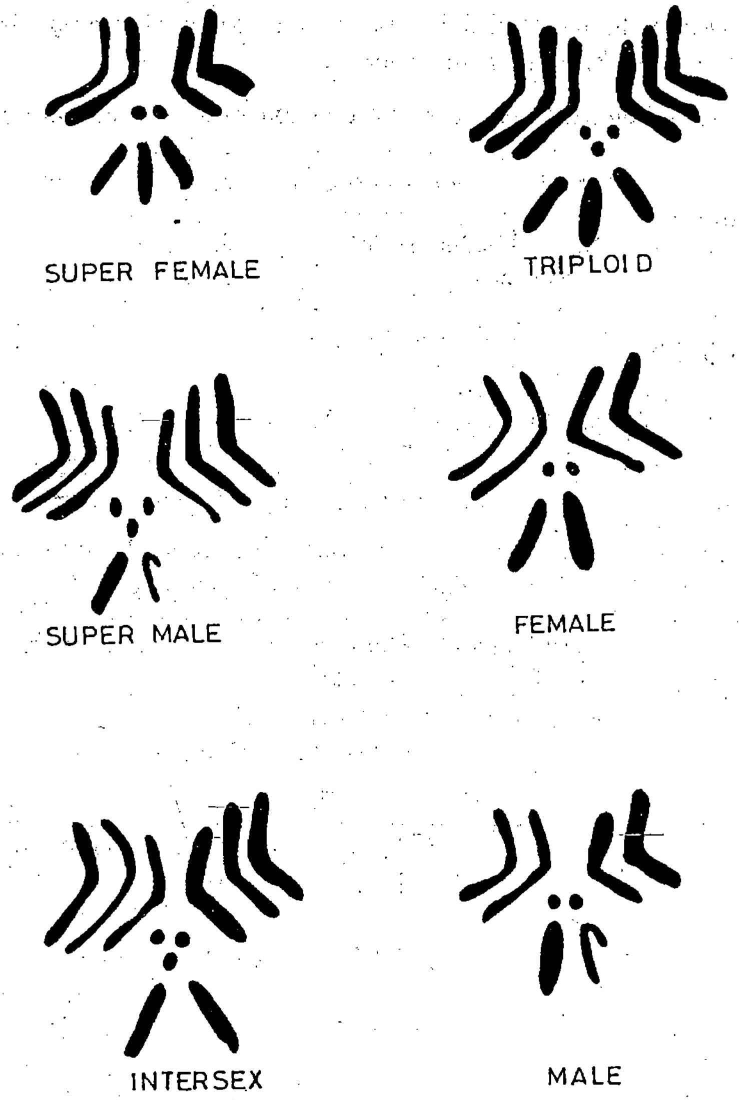
The X chromosomes as the determiner of sex was discovered by McClung in 1902. McClung studied the spermatogenesis in the grasshopper and discovered in the stages preceding the formation of sperms, all the chromosomes of various sizes and shapes were paired with a lone exception, which was seen without a mate. This lone chromosome was called as the "Odd chromosome" or the 'X' chromosome. In later stages of spermatogenesis when all the paired chromosomes in the reduction division separated following synapsis, only half the gametes (mature sperm) contained the "odd chromosome", while the other half contained no odd chromosome. McClung's discovery gave a stimulation to the study of sex determination by cytogenetic studies. Stenes and Wilson independently studied spermatogenesis on various species of insects. They found in each of a male, the 'odd' chromosome and in the female two 'odd' chromosomes. The male had a partner different in shape and size called the Y chromosome, males are XY and females are XX in the sex chromosome make up. The other chromosomes in the individual are called the autosomes.

The 2A +XX genotype indicates the female sex and the 2A +XY indicates the male sex. This suggests the possible explanation that the 'Y' chromosome in the male is that which determines the sex. But the absence of the 'Y' chromosome in the Drosophila resulting in a individual 2A +X is a perfect phenotypic male but it is sterile thus indicating the presence of the Y chromosome is necessary for fertility. In addition the presence of a Y chromosome in female (XXY) is a fertile female thereby showing 'Y' has no effect in the presence of XX.

Genic balance theory:

Studies on the sex determination of Drosophila by Bridges reveal not only the sex chromosomes but also the autosomes that have a bearing on sex determination. This is known as the "Genic balance theory of sex determination".

Bridges in 1921 indicated that in Drosophila the simple presence of XX or XY is not enough to fully explain the determination of sex in these insects. He found a balance exist between the sex chromosomes on one hand and the autosomes on the other. A Drosophila with the X chromosomes and with three sets of autosomes (3A +XX) is not a female but an intersex. An intersex is a sterile individual having the phenotypic characters intermediate between male and female. The only difference between the intersex and the normal female is that the intersex has got one more set of autosomes. This indicates that not only the X chromosomes but also the autosomes have bearing on sex determination. The autosomes carry certain male determining strength. The female determining strength of each X chromosome is 1.5 and the each haploid set of autosomes has a male determining strength of one. The normal female is 2A+XX. The male determining strength is 2x1 = 2. The female determining strength is 1.5 x 2 = 3.0. The ratio of the male tendency to female tendency is 2:3. In the normal male the ratio of male tendency to female tendency is 2:1.5. In the



Genic Balance Theory of Drosophila Melanogaster normal female the female strength is more and in the normal male the male strength is more. In the intersex the male strength to female strength is 3:3, and hence it is neither a male nor a female.

The genic balance theory can also be expressed as a sex ratio or sex index:

$$\text{Sex ratio} = \frac{\text{The No. of x chromosome}}{\text{No. of Haploid set of autosomes}} = \frac{X}{A}$$

In a normal female $\frac{X}{A}$ ratio is $\frac{2}{2} = 1$

In a normal male $\frac{X}{A}$ ratio is $\frac{1}{2} = 0.5$

If the $\frac{X}{A}$ ratio is more than one it is a super female and $\frac{X}{A}$ ratio is less than 0.5 it is a super male. X/A ratio if between 0.5 and 1.0 are intersexes. The following table gives the different sex types:

No.of X chromosomes	No.of Haploid sets of autosomes	The ratio of male tendencies to female tendency	$\frac{X}{A}$ Ratio	Sex type
3	2	2:4.5	1.5	Super female
4	4	4:6 (2:3)	1.0	Female (Tetraploid)
3	3	3:4.5 (2.3)	1.0	Female (Triploid)
2	2	2:3	1.0	Female (Diploid)
3	4	4:4.5	0.75	Intersex
2	3	3:3	0.66	Intersex
1	2	2:1.5	0.5	Male
1	3	3:1.5	0.33	Super male

SEX DETERMINATION AND SEX DIFFERENTIATION IN MAMMALS

Sex determination is mainly by the chromosome complement namely an individual XX is female and an individual XY is a male. But sex differentiation consists of the differentiation of the indifferent gonad by the chromosomal complement and sex differentiation of the extra gonadal structures by hormones.

Every individual XX or XY at the beginning of development of sexual organisation possess (i) a pair of undifferentiated gonad (ii) a pair of Mullerian ducts and a pair of Wolffian ducts and (iii) the urogenital sinus, the structures concerned in later development of the secondary gonadal characters of both sexes.

The primary sex determination is by the chromosomal mechanism and is concerned with the differentiating of the undifferentiated gonad into the formation of a testes or ovary. If the chromosomal sex is XX the gonad is differentiated into an ovary and if it is XY gonad is differentiated into a testis.

If the gonad becomes an ovary the Wolffian ducts regress. The Mullerian ducts transform into Fallopian tubes, uterus and the upper third of a vagina. The urogential sinus and genital tubercle transformed into the vulva and clitoris.

If the gonad becomes testes the Mullerian ducts regress. The Wolffian duct differentiate into epididymis, vas differentia and seminal vesicles. The urogential sinus and genital tubercle results in the formation of scrotum, penis and urethra.

INTERSEXES

An intersex is a sterile individual having the characteristic intermediate between the male and female. Intersexes arise when there is a conflict between the four criteria of sex namely the chromosomal sex, the sex of the gonad, the external apparent sex and the physiological sex.

Intersexuality of Farm Animals:

Any classification of intersexuality must take into consideration the nuclear sex, type of gonadal tissue, extragenital structures and sex behaviour of the individual. Intersexuality can be classified into:

1. Freemartinisms
2. True hermaphroditism
3. Pseudo hermaphroditism
4. Behavioural forms.

FREEMARTINISM

Twins in cattle may exist as both normal males and both normal females. One male and one female both normal, and one normal male and the other externally a female with an abnormal reproductive system is known as freemartin. Most of the twins in cattle are binovular because two corporalutea are formed and two ova are concerned in the pregnancy. The two fertilised eggs pass into the uterus and become attached to the uterine mucosa. If the two ova have been released from the same ovary, the zygotes developed in one and the same uterine horn. As the egg increases in size the embryonic membrane is formed in the two foetuses. The membranes tend to adhere in some cases. If such fusion occur, an anastomosis of the blood vessels can occur so that a common vascular intercommunication is established between the two foetuses. Thus the situation arises in which the sex hormones of each developing individual is at liberty to pass into the tissues of its cotwin. If the two twins are of the same sex nothing abnormal happens, but if the twins are of the opposite sexes and fusion of the chorion occurs, a vascular communication is developed. The sex differentiation of both the individual will be directed by that sex hormone which is formed earlier. The male sex hormone is liberated before that of the female. The female twin will therefore purpose her sex differentiation under the direction of the male sex hormone from the cotwin and will therefore come to possess more or less completely the organization of the male. The external genetalia are of the female pattern, the internal organ of reproduction are more or less completely male.

The bovine freemartins are undersized. Gonads are intraabdominal and consists of ovary or ovotestis. The genital tract is intersexual. The external genitalia are of the female type. Frequently the clitoris and the tuft of hair at the vental commissure of the vulva are enlarged. The external genitalia lead into a short vagina. The mammary glands although under developed can be palpable. Teats shorter than those in normal heifers. Freemartins do not experience heat.

TRUE HERMAPHRODITISM

True hermaphrodites are common in pigs and rare in cattle and horses. In the true hermaphrodites, one gonad may be an ovary and the other a testis, or both gonads may be ovatestis. The testis may be located subcutaneously in the inguinal region or within the abdominal cavity.

Cattle:

True hermaphorodites have been reared as heifers because they show female external genitala. Testes are either rudimentary or poorly developed. The vagina and cervix are usually underdeveloped.

Goats:

The degree of intersexuality varies from female type to the predominently male type. In the female type, the external appearance may be that of a normal female but the animal may have seminal vesicles and vasa deferentia. In the predominantly male type, the penis and descended testis are poorly developed and the Mullerian duct survives along with the vas deferens. True hermaphroditism is a hereditary trait and is more frequent in certain breeds of goat. For example Sanon, Toggenburg, Angora and some milch breeds. The anomaly has been traced to the action of a recessive autosomal gene. True hermophroditism is hereditary and shows close to 100% linkage to polledness, it can be eliminated by using horned breeding stock.

Pigs:

True hermaphroditism in pigs is common. The presence of both ovarian and testicular tissue seems to account sufficiently for the intersexual character of the accessory organs in this type of intersex. Male organs are represented by testis, epididymis and vas deference. The female reproductive tract is represented by fallopian tubes, uterine horns, cervix and vagina. Externally normal labia, enlarged clitoris with the opening on its dorsal surface and a normal prepuce are present, the teats and mammary glands are lacking.

PSEUDOHERMAPHRODITISM

More common in the male than in female. Male hermaphrodites have undescended testes and varying combinations of male and female genital structures so that the external genitalia are often quite ambiguous.

Cattle:

Several cases of male pseudohermaphroditism in cattle have been reported. These animals resemble steers and do not exhibit manifestation of sexual desire. The external genetalia are always of distinct female character with smaller than normal vulva, clitoris and mammary gland. The scrotum may or may not be present, and if present is usually empty. Rudimentary male external genetalia may include a prepuce 2 to 3" below the location of the vulva, and a poorly developed penis. The internal genetalia comprise male accessory structures e.g., vas deferens, seminal vesicles and sometimes a prostate gland. The majority of male pseudohermaphrodites were born as twins to normal calves.

Goats:

In male pseudohermaphrodites, the sexual organs were combinations of poorly developed male and female organs.

Pigs:

Male pseudohermaphroditism is of frequent occurrence. Animals have the external appearance of gilts, but with the onset of puberty a change in external appearance and behaviour is evident. Such animals behave like boars and even mount gilts in oestrus. Enlargement of the clitoris distorts the contours of the vulve. Testes are located either intra-abdominally or subcutaneously just below the vulva. Rarely is a scrotum present. An epididymis, vas deferens, prostate glands and seminal vesicles are frequently present. Usually uterus is well developed. The cervix is poorly developed and the vagina is either small or absent.

BEHAVIOURAL INTERSEXHALITY

Nymphomania and adrenal virilism are typical syndromes involving behavioural intersexuality in cattle.

Sex Reversal:

Sex reversal is meant the transformation of sexual characterisation of an individual from that which is normal in one functional sex to that which is normal in the opposite sex. It is the process by which an individual that functioned as a male or female as the case may be, becomes transformed into one that can function as a male or female as the case may be. In other words, the reversal is complete. Incomplete sex reversal is what is known as an intersex.

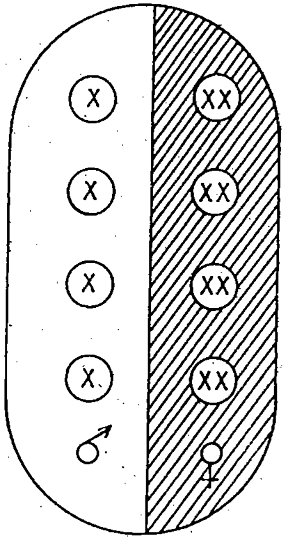
For sex reversal to be complete it is necessary for that component structure of the sex equipment must be capable of transformation or replacement that is, one kind of gonadic structure must be replaced by the other. The accessory sexual apparatus, the external organs of reproduction, the rest of the secondary gonadic characters must be capable of becoming remodelled or replaced. Sex reversal cannot occur in mature individual, the external and internal genetalia of which are fashioned early on its life history and therefore loose all embryonic plasticity and become unresponsive to any stimulus. Sex reversal also cannot occur in any case in which the differences between male and female architecture are based upon the differential development of the two sets of structures, one of which in either sex undergoes complete atrophy. Sex reversal also cannot occur in cases in which the sexual dimorphism involves a differential mode of development of one and the same set of structures i.e. uro genetal sinus or tubercle in mammals.

Sex Reversal in Mammals:

Sex reversal seems to be incompletable with the peculiarities of uterine development possessed by mammals. Further the fact that a complex genetal apparatus differentiate early on the embryonic life in mammals makes sex reversal during the late embryonic stages impossible because once differentiation of sex has proceeded very far in either direction there can be no retracing of the steps and hence sex reversal cannot be expected in mammals.

Sex Reversal in Birds:

It is not impossible theoretically because the external genetalia in the



Bilateral Gynander in Drosophila

A fly with male tissue on one side and female tissue on the other side

two sexes are very similar and because experimentation has shown that the functional gonads play an all important role in sexual differentiation. It is being possible to masculinise a female and feminise a male by means of appropriate gonad implantation. In order that sex reversal may occur in birds two conditions are necessary.

i) Gonadectomy through disease.

ii) Development of new sex cords.

The development of new sexcords can occur only in mature birds and it can develop thoroughly only into testicular tissues. Hence complete sex reversal may be possible in the case of the hen, i.e. the hen can transform into a cock and the cock can never become a functional hen. Crew (1923) described a typical case of complete sex reversal in Buff orpingor hen. The hen was laying eggs upto 3 1/2 years old, suddenly ceased to lay eggs. It gradually developed comb, wattle, and spur and the pattern of cock feathers. It becomes aggressive like a cock and mounted females. It was placed with a virgin Buff orphington, pullet and eggs were collected and incubated and two chicks were hatched out. The sex reversal is complete here, i.e. a functional female becoming a functional male.

GYNANDROMORPHISM

An intersexual condition due to a regional disharmony in the distribution of sex chromosomes. Gynandromorphs will have male tissue in one part of the body and female tissue on another part. It has been seen in insects in which sex hormones are absent, the control of sex is by the chromosomal complement which makes it possible to get individual with female tissue on one part of the body and male tissue on the other. The most commonly seen is the bilateral type of gynanders in which one side is fully male and another fully female. In some it may be antero-posterior and in some it may be a patchwork of female and male tissue called as sex mosaic. "Mosaicism" is a term used to describe the condition where an individual cell consists of two or more populations each with different chromosomal complement.

In Drosophila, gynandromorphs arise from zygotes that have originally two X chromosomes namely female. The gynanders start their life cycle as females with a normal pair of X chromosomes. In the first mitotic division however, it is possible for one of the X chromosomes to be lost in the cytoplasm and failed to be included in the nucleus of one of the two cells formed. Thus in the two cell stage one normal female cell is matched with a cell having only one X chromosome, a fact which makes this cell to form male tissue. As these two cells continue to divide, the bilateral type of gynanders is produced.

The antero-posterior gynandromorphism is noticed in the wasp habrobrocan. A male head with female abdomen and a female head with male abdomen is seen.

Gynandromorphs are impossible in the vertebrates because sex hormones are distributed all over the body. Sex hormones in vertebrate tend distribution of sex all over the body.

PARTHENOGENESIS

The development of an unfertilised egg in an individual is known as parthenogenesis. This is a method of sex determination in the order hymenoptera like honey bees, ants and wasps.

A classical example can be given in honey bees. Only female honey bees namely the queen-and workers come from fertilised eggs, the unfertilised eggs are developed into males (Dreones). This finding has been substantiated both genetically and cytologically. The male honey bee has got 16 chromosomes and the female has 32 chromosomes (16 pairs). In this case the sex depends upon whether or not fertilisation occur.

Studies in spermatogenesis in bees revealed the fact that the spermatogonia possess the haploid number of chromosomes instead of the diploid number (32). In order therefore this haploid number may not be further reduced in meiosis, only one division of chromatin takes place in spermatogenesis. In the primary spermatocyte division all the 16 chromosomes pass into a secondary spermatocyte and only a minute degenerative non-chromatic globule is formed in the other spindle. The secondary spermatocyte divides into one spermatid and the other being small degenerates, thus the sperms carry the haploid number of chromosome.

EXERCISE

1. Describe the three mechanisms of sex determination.
2. Discuss on the following theories of sex determination.
   a) xy chromosome method
   b) Genic balance theory
3. Write short notes on
   a) Intersex
   b) Freemartinism
   c) Hermaphroditism
   d) Sex reversal
   e) Gynandromorphism
   f) Parthenogenesis
4. In Drosophila, the ratio between the number of x chromosomes and the number of sets of autosomes A is called the sex index. Calculate the sex index and phenotype in the following individuals :
   a) AAX b) AAXXY c) AAAXX d) AAXX e) AAXXX
   f) AAAXXX g) AAY
5. Distinguish between
   a) Sex determination and sex differentiation
   b) Heterogametic and homogametic sex
   c) Autosomes and sex chromosomes
   d) Freemartin and pseudohermaphrodite
   e) Intersex and Gynandromorphs
   f) Sex mosaics and Gynandromorphs

Molecular Genetics

Chapter 20

Gene Mutations

Mutation is a change in the gene, potentially capable of being transmitted.

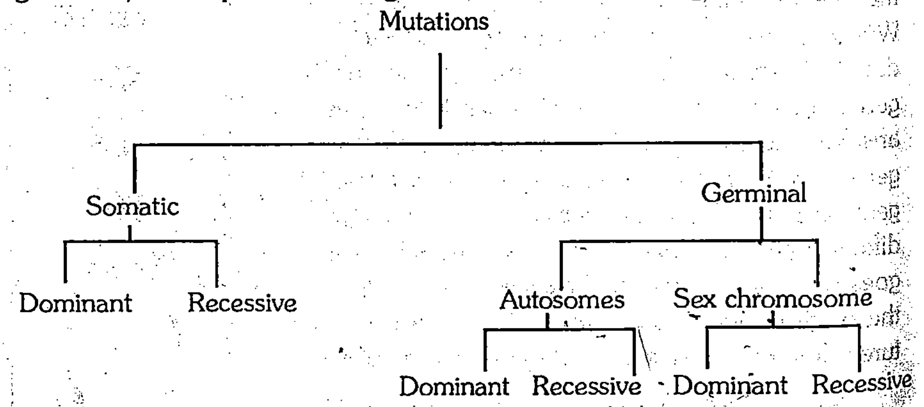
The genotype of an individual remain constant from fertilisation to birth unto death. Stricktly speaking it is not correct. The adult person does not have the same gene as what he had as an infant. What the adult has are only copies of these genes. Similarly children do not have the genes of their parent but only copies of the parental genes. When a chromosome divides it gives rise to two daughter chromosome with similar genes. The genes must have reproduced themselves. Genes arise only from genes. Heredity is due to accurate gene reproduction. Occasionally, the process of gene reproduction goes wrong i.e. a copy of a gene differs from the original gene. The modified gene goes on reproducing its changed structure just as the original one. This change in the gene structure is known as Gene mutation or Point mutation.

First mutation in animals were observed in 1791 by Sethwright. He found in his flock of sheep, a sudden birth of a short legged lamb. He reared this lamb and bred from it a variety of short legged sheep and was instrumental in bringing out this as a separate breed, ANCON, through judicious breeding approaches. This breed became extinct, but later another short legged sheep occurred in a flock of sheep in the Norwegian farm representing the new occurrence of same mutation. From this a new strain of short legged sheep has been bred. Similarly the hornless cattle appeared in the breeds of horned cattle. As per Darwin's theory of evolution, the new hereditary type recorded in plants and animals are called 'sports'. They are nothing but mutations which gave rise to new variety of breeds. In late 19th century, Devries putforth the theory of mutation, that hereditary is due to changes which have been large and discontinuous. Devries called these large effects as mutations. He recorded mutations in the evening Primroses.

The first scientific study of mutation was reported in 1910 by T.H. Morgan. He obtained white eyed Drosophila in a culture of red eyed flies. Later in a period of 16 years, nearly 500 mutations affecting colour of the body, colour of the eye, length of wing etc. were reported.

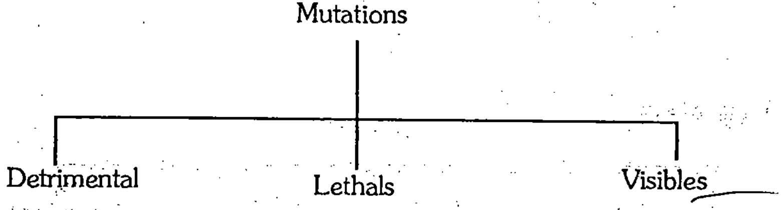
Classification:

Based on the location mutation may be somatic or germinal. If it occurs in somatic cell it shall be restricted to the individual, whereas if it occurs in germ cell, it is capable of being transmitted to the next generation.



Mutations breed true. If it is dominant it can express itself immediately. Likewise a sex linked mutation even recessive can readily express in the heterogametic sex since expression is possible in them in single dose. Recessive mutations take generations to appear phenotypically.

Mutation may be classified based on its effect as detrimental, lethal and visibles.



Detrimentals are more common and of 80% of the mutations generally. They have no visible effect but cause some decrease in viability of the organisms possessing them.

Lethals occur over 19% of mutation. They have a physiological effect so extreme as to cause the death of organism possessing it.

Visible mutation are less in occurrence. They are less than 1% of mutations. They cause clearly detectable visible phenotypic effect.

Mutation can occur in both directions. Based on direction it may be forward mutation or backward or reverse mutation.

Wild type Mutant type

$A \longrightarrow a$

$A \longleftarrow a$

Backward

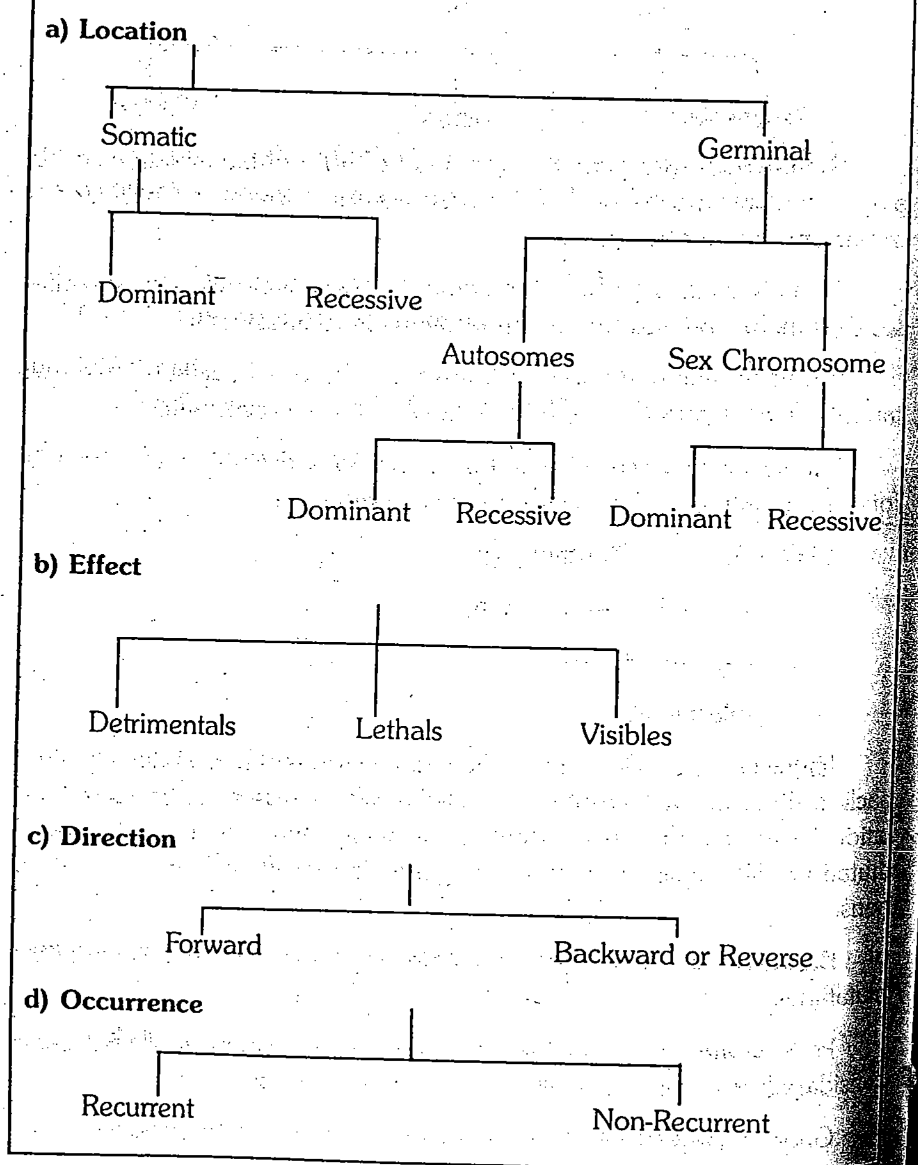
The wild type allele can mutate to a new mutant type and again revert back to the original wild type. This is known as reverse or backward mutation. The rate of forward mutation is always higher than the backward mutation. Backward mutations are generally one tenth of forward mutations.

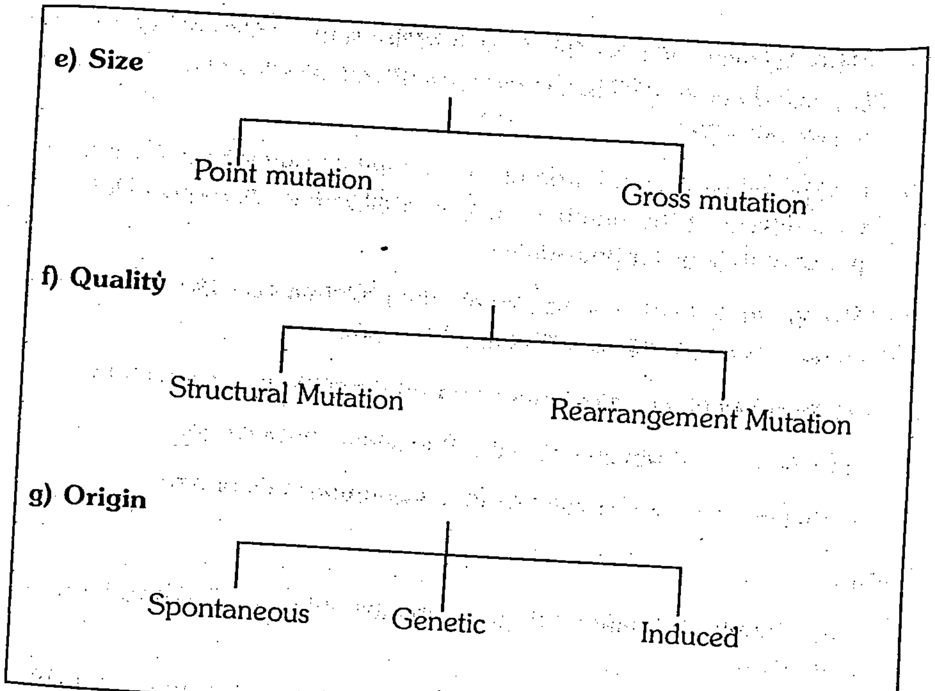
Based on size mutations can be classified into point mutation and gross mutations.

Point mutation - a change in a very small segment of DNA; usually considered to involve a single nucleotide or nucleotide pair.

Gross mutations - changes involving more than one nucleotide pair;

Classifications of Mutation


a) Location
Somatic
Germinal
Dominant
Recessive
Autosomes
Sex Chromosome
Dominant
Recessive
Dominant
Recessive
b) Effect
Detrimentals
Lethals
Visibles
c) Direction
Forward
Backward or Reverse
d) Occurrence
Recurrent
Non-Recurrent


e) Size
Point mutation
Gross mutation
f) Quality
Structural Mutation
Rearrangement Mutation
g) Origin
Spontaneous
Genetic
Induced

may involve the entire gene, the entire chromosome, or sets of chromosomes (polyploidy).

With respect to quality mutations may be structural or rearrangement.

a) Structural mutations - changes in the nucleotide content of the gene.

1) Substitution mutations - substitution of one nucleotide for another. a) Transition mutations substitute one purine for another or one pyrimidine for another. b) Transversion mutations substitute a purine for a pyrimidine or vice versa.

2) Deletion mutation - loss of some portion of a gene.

3) Insertion mutants - addition of one or more extra nucleotides to a gene.

b) Rearrangement mutations - changing the location of a gene within the genome often leads to "position effects".

1) Within a gene - two mutations within the same functional gene can produce different effects, depending on whether they occur in the cis or trans position.

2) Number of genes per chromosome - different phenotypic effects can be produced if the numbers of gene replicas are non-equivalent on the homologous chromosomes.

3) Moving the gene locus may create new phenotypes, especially when the gene is relocated near heterochromatin.

   a) Translocations - movement to a non-homologus chromosome.

   b) Inversions - movement within the same chromosome.

Mutations may be classified as follows based on its origin.

Origin:

a) Spontaneous mutation - origin is unknown; often called "background mutation".

b) Genetic control - the mutability of some genes is known to be influenced by other "mutator genes".

1. Specific mutators - effects limited to one locus.
2. Non-specific mutators - simultaneously affects many loci.

c) Induced mutations - through exposure to abnormal environments such as:

1. Ionizing radiations - changes in chemical valence through the ejection of electrons are produced by protons, neutrons, or by alpha, beta, gamma or X-rays.
2. Non-ionizing radiations - raise the energy levels of atoms (excitation), rendering them less stable (e.g. ultraviolet radiation, heat); UV often produces thymine dimers, i.e. bonding between thymines on the same strand.
3. Chemical mutagens - are chemical substances which increase the mutability of genes.

a) Copy errors - mutants arising during DNA replication. e.g. base analogue mutagens which are chemically similar to the nucleic acid bases, may be incorporated by mistake; acridine causes single base additions or deletions possibly by intercalation between two sequential bases.

b) Direct gene change - produced in non-replicating DNA, e.g. nitrous acid by deamination directly converts adenine to hypoxanthine and cytosine to uracil.

Characteristics of mutation

The known facts may be arranged in a series of statements.

1. Many genes are stable.

   Muller estimated the mean life of a gene in Drosophila to be 10,000 years. Muller has estimated that about one in a million genes appear to mutate at a particular locus in Drosophila. Rate of natural mutation is very low.

2. Different genes have different rates of mutations.
3. Mutations may occur at any point in the life history of an organism. They may occur in somatic or gametic tissue but more frequently during maturation of the germ cells.
4. The rate of mutation in various genes may vary in different tissues of different stages of development.
5. A mutation is a change in a gene, and not the loss of the gene.

   When A is mutated to a, a can also be mutated to A.

   $A \longrightarrow a$

   $A \longleftarrow a$

6. Mutation can occur in both directions. The rate of forward mutation is always higher than the backward mutation.
7. Reverse mutations are 1/10th of forward mutations.

8. More than one change may occur in a gene resulting in multiple alleles.

   (Eg) C - for colour in rabbit

   converts into c^{ch} - Chinchilla

   c^{h} - Himalayan

9. Multiple allelic mutations of a gene is not always in the same direction.

10. The direction of mutation is however preferential occurring more often in same direction than others.

    (Eg) Rose to purple in flower colours.

11. The mutability and preferential direction may themselves become changed through mutation.

12. The changes in a gene is basically in its chemical structure.

13. Gross mutations are usually harmful to organisms.

14. Mutations are generally recessive to wild type, but dominant mutations may also occur.

15. Mutations are sudden in occurrence.

16. Two alleles of a gene in an organism mutate independently of each other.

17. Mutations do not ordinarily occur in more than one gene at a time.

18. Mutations with slight effects are more common of all mutations.

19. Mutations with no visible effects are most common and may be unnoticed.

20. Radiation (X-rays, radium rays, ultraviolet rays, heat rays) may greatly increase the natural mutation rate. Muller was awarded Nobel prize in 1946 for his finding on the effect of radiation on the production of mutations.

21. Certain chemicals may also cause mutation. Sulphur mustard, Nitrogen mustard, Allyl isothiocyanate, Ethyl urethane, Phenol Methylcholanthrene, Dibenzanthrene, Benzyprene etc. are mutagenic substances.

    Chemical mutagens produce gene mutation as well as chromosomal mutations.

22. Temperature can be a factor in the induction of mutation in some organisms. Muller observed that exposing the larva of Drosophila to a high temperature (just below that killing) the mutation rate was increased to six times than the normal rate.

Naturally occurring mutations include all changes in DNA sequence. These may be from simple switches of one base for another to completely destroying the functions of the gene. These drastic changes are called mull mutations, which may include base switches, insertions, deletions of a base, or gross rearrangement of chromosome structure.

EXERCISE

1. Define mutation and discuss on the reasons for its occurrence.
2. Classify mutations on different aspects.
3. Give an account on the salient features of mutation
4. Why are sex linked lethal mutations easier to detect than autosomal lethals?
5. How will you determine whether a mutation is a somatic one or is due to a genic change?
6. A character not present in the parents appears in the progeny. How will you decide whether the new character is due to a mutation or recombination?
7. Adenine and cytosine are susceptible to the same mutagen whereas thymine and guanine are susceptible to the same mutagen. Why?

Chapter 21

Chemical Basis of Heredity

Chemically genes are made up of Deoxyribose Nucleic Acid (DNA).

In 1869, Frederick Meicher, a Swiss biochemist isolated the substance called 'Nuclein' from the pus cells. The nuclein had the properties of an acid and hence called Nucleic acid. Watson and Crick of England, and Mirsky and Ris of America analysed chromosomes and observed that they are made up of nucleo proteins.

Nucleoproteins consists of proteins combined with nuclear acid. The nuclear acids have high molecular weight and are highly compounded i.e. Polymerised. Two groups of nucleic acids are known for taking into account their names from the sugar which is an essential part of it.

1. Ribose nucleic acid - RNA - confined to cytoplasm.
2. Deoxyribose nucleic acid - DNA is confined to nucleus.

Degradation of these nucleic acids by enzymes or acids reveals that they are made up of three essential components.

1) Pentose sugar

2) Phosphate and

3) Nitrogen base - Purine and Pyrimidine

The sugar + base (Purine or Pyrimidine) gives Nucleoside.

The sugar + base + Phosphate combinations give rise to Nucleotide.

The components of DNA and RNA are differentiated in the Table below:

Components	**RNA**	**DNA**
Sugar	Ribose	De-oxyribose
Phosphate	Phosphate	Phosphate
Purines	Adenine & Guanine	Adenine & Guanine
Pyrimidine	Uracil & Cytosine	Thymine & Cytosine
Structure	Single strand polymer	Double strand polymer

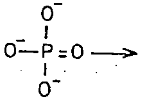
A molecule of nucleotide consists of one molecule of sugar, one molecule of base and one molecule of phosphoric acid.

Chemical structure of DNA

DNA is a polymer built up of a long chain of many mononucleotides. Each mononucleotide in DNA consists of one molecule of sugar deoxyribose, one molecule of phosphoric acid and one molecule of base (Purine or Pyrimidine). The sugar of one nucleotide may be attached to the phosphate of another and so on in a regular sequence forming long molecules. The alternating sugar and phosphate group form a long and straight chain with the purine and pyramidine projecting out from the chains at right angles.

The DNA molecule consists of two chains running parallel to each other

The Structural Formulas for the Constituents of Nucleic Acids

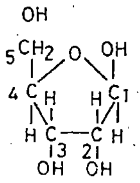


$O^- - P(=O)(O^-) - O^-$

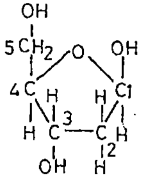
→ A PHOSPHATE GROUP

A Five Carbon Sugar of Pentose

IN RNA:RIBOSE

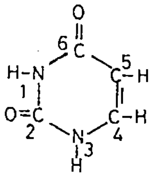


IN DNA:2-DEOXY RIBOSE

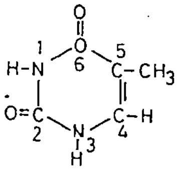


A Cyclic Nitrogen Containing Bases

IN RNA ONLY
URACIL
(2,6-Oxy Pyrimidine)

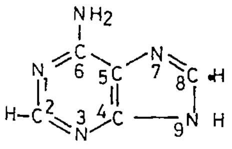


IN DNA ONLY
THYMINE
(2,6 Oxy-5-methyl Pyrimidine)

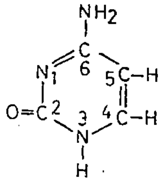


In Both RNA and DNA

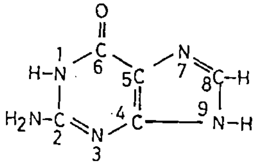
ADENINE
(6-Amino purine)



CYTOSINE
(6-Amino-2-Oxy Pyrimidine)

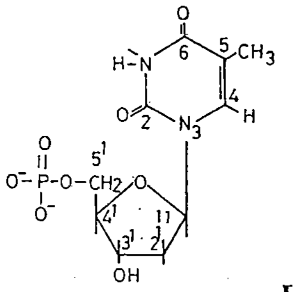


GUANINE
(2-Amino-6-oxy purine)

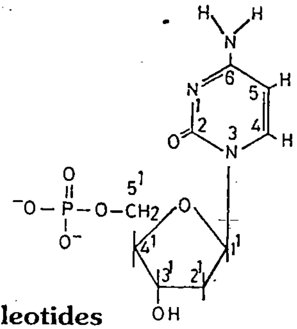


Pyrimidine Nucleotides

Deoxy Thymidine Mono Phosphate, dTMP

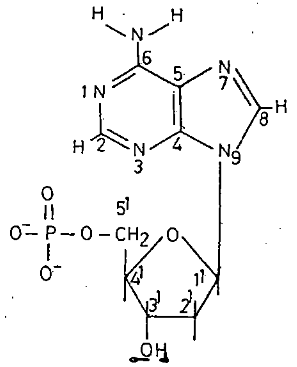


Deoxy Cytidine Mono Phosphate, dcMP

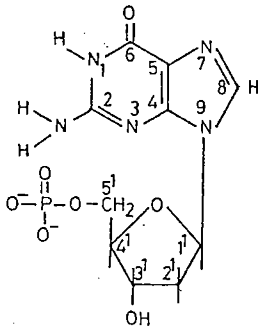


Purine Nucleotides

Deoxy Adenosine Mono Phosphate, dAMP



Deoxy Guanosine Mono Phosphate, dGMP



The four common Deoxyribonucleotides of DNA

RNA contains ribonucleotides with the pyrimidines uracil and cytosine and the purines adenine and guanine

कृपया रखा पारुन
पुस्तक खराब करु न

but oriented in opposite direction. Each purine of one chain is attached to the pyrimidine of another chain by the bonds forming a series of short cross chains between the two long chains. DNA molecule can be compared to the ladder made of chains. Each of the two lengthwise chains of the ladder is made up of the units of deoxyribose and phosphate alternating one with the other along the length of the chain. The cross chains are fairly straight and quiet regular. The two lengthwise chains run in opposite direction and there is no distinctive top or bottom to DNA molecule. Each short chain or the rung of the ladder connects deoxyribose of one chain with the deoxyribose of other chain. Each cross chain consists of a purine and a pyrimidine bonded together by means of hydrogen bonds.

The purine Adenine always combines with the pyrimidine Thymine and the purine Guanine always combines with the pyrimidine Cytosine.

Watson and Crick Model of DNA:

According to Watson and Crick, DNA molecule is composed of two polynucleopeptide chains. The two chains are coiled around a common axis to form a helix which is made up of proteins. The two chains are joined together by H_2 bonds between the two nucleotide bases. The phosphates and sugar groups are at the outside of the helix. The bases are on inside. The distance from the axis of the helix to the phosphate is 10A (Angstron) unit. The unique features of the structure rests on the arrangement of the bases. The bases are arranged perpendicular to the fibre axis and are joined together in specific pairs. As per the structure the adenine specifically pairs with thymine and guanine specifically with cytosine. The chains are complementary i.e. once the sequence of the bases in one chain is known the necessity for specific pairing determines the sequence of bases in the second chain. This features of complementary makes the structure of DNA of great interest.

Specific pairing of bases:

Purine with double rings are larger than the pyrimidines with single ring. If two purines are paired their dimensions are too great to fit the constant diameter of the helix while the dimensions are too small, if two pyrimidines are paired.

Kinds of DNA - Kinds of Gene

To account for all kinds of genes there are enough kinds of DNA molecules, which may be listed as follows:

1. Relative number of Adenine-thymine and guanine-cytosine cross chains.
2. The order in which these cross chains occur.
3. The direction in which each cross chain is attached to the long chain.

Because the DNA molecules are very large the permutations and combinations of them are tremendous.

Fine structure of the gene:

The relationship of the genes to the long double coils of the DNA reveals the gene as a unit of the recombination; a unit of mutation and a unit of function.

The unit of recombination is called 'Recon' which consists of a pair of nucleotide or at the most two pairs of nucleotide.

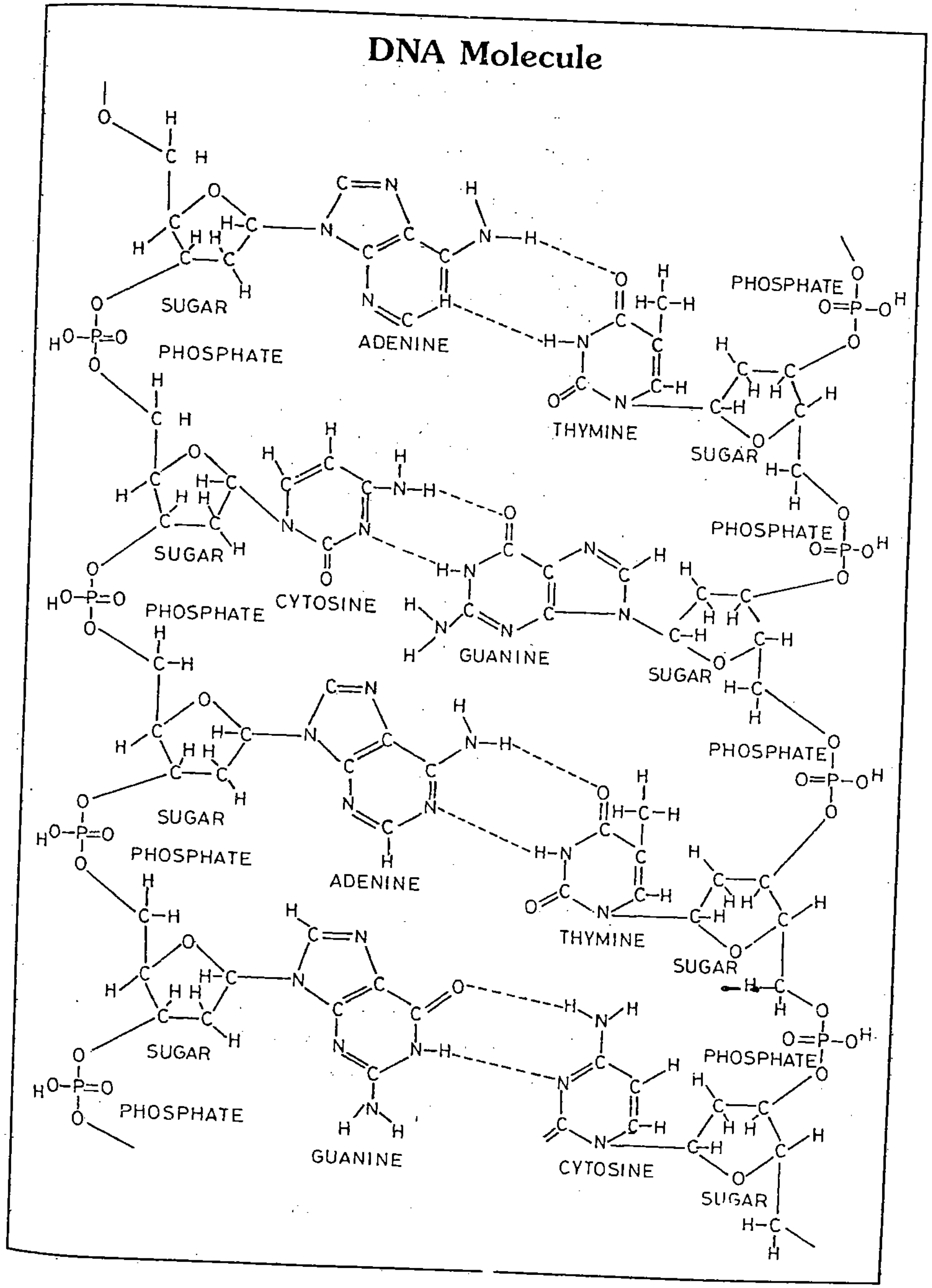
The unit of mutation is called 'Muton'. It is the smallest part which when altered can give rise to a mutation. The alteration of a single nucleotide results in mutation.

The unit of function is called "Cistron". This is commonly called as gene. The cistron consists of about 500 nucleotide pairs. Some may have even as long as 30,000 nucleotide pairs.

Evidence for the DNA to be the true genetic material

Indirect evidence	Direct evidence
The correlation between properties of genetic material and the DNA	by conducting experiments

DNA Molecule


SUGAR
PHOSPHATE
ADENINE
THYMINE
SUGAR
PHOSPHATE
SUGAR
PHOSPHATE
CYTOSINE
GUANINE
SUGAR
PHOSPHATE
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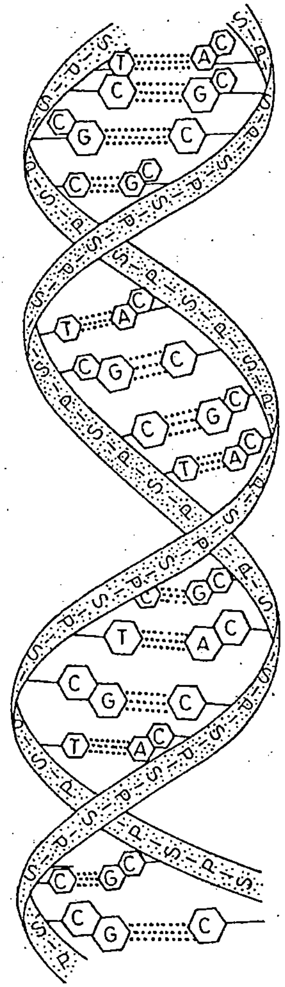
CYTOSINE
SUGAR



Watson-Crick Double Helix Model of Structure of DNA

A - Adenine
B - Thymine
G - Guanine
C - Cytosine
S - Deoxyribose Sugar
P - Phosphate

Indirect evidences:

1. There is a correlation between the location of the gene and the location of DNA, both are located on the chromosomes which form the part of the nucleus.

2. The number of chromosomes in a somatic cell is always constant. The amount of DNA for somatic cells is also constant and the amount is not affected even in the metabolic state of cells. The haploid cells have only one set of chromosome. The amount of DNA in the sperm and egg is also, half the amount of DNA present in somatic cell. Further in the polyploid cell the amount of DNA in multiples of halpolid amount.

Cattle:

Thymus = 6.6 mgm x 10^{-9}
Liver cells = 6.4 mgm x 10^{-9}
Pancreas = 6.9 mgm x 10^{-9}
Sperm = 3.3 mgm x 10^{-9}

Fowl:

RBC = 2.34 MG X 10^{-9}
Liver = 2.39 mg x 10^{-9}
Sperm = 1.26 mg x 10^{-9}

Genes are carried on the chromosomes and there is a constant number of chromosome per cell and also the DNA content per cell is also constant. This shows there is a correlation between the genes and DNA. The number of purines are always equal to the number of pyrimidines ($A+G = T+C$) ($A=T$; $G=C$:) $A+T/G+C$ = The base ratio. This base ratio is constant for a given species.

3. The ultraviolet radiation is definitely mutogenic. The action spectrum of ultraviolet radiation is about 2500-2600 A and the absorption spectrum of the nucleic acid is about 2600 A. This shows that there is a

definite correlation between the absorption spectrum of nucleic acid and the action spectrum for mutation. This observation has been interpreted as indicating that the DNA is the target molecule and the changes resulting in nucleic acids have resulted in mutation.

Direct evidences:

Studies with micro organisms on the two phenomenon "Transformation" and "Transduction" give a direct evidence to say that the DNA is the genetic material.

Transformation:

Studied in the bacteria Pneumococci - Two types of pneumococcus were known: (1) 'S' type which has got a smooth capsulevirulent type and (2) R type has no capsuleavirulent strain. The 'S' cells injected to mice cause septicemia and death R cells do not. The R cells mixed with heat killed 'S' cells causes septicemia. That is the R cells which is uncapsulated becomes changed into 'S' cells which is capsulated and cause septicemia. In other words avirulent becomes virulent strain. The particular substance of the killed 'S' cells responsible for transformation was proved to be DNA by Avery, Macleod and MacCarty (1944).

In transformation the genetic material in the form of particles of DNA can penetrate bacterial cells and can become incorporated into the genetic material of the cell, possibly replacing homologous genetic material on a host chromosome.

Transduction:

Studied in salmonella and E. coli. The replacement of DNA from one bacterium into another takes place by bacteriophage which penetrates the host cell. The bacteriophages are virus which attack bacteria. Viruses are organism very much smaller than bacteria which can invade living cells and can reproduce within the cells. Phages are composed of both protein and DNA, but only the DNA enters the host cells. The protein left outside is a further indication that although chromosomes are composed of nucleoprotein, the genetic material itself may be DNA.

EXERCISE

1. Differentiate between DNA and RNA.
2. Draw the double helix model of DNA.
3. Give an account on the kinds of genes.
4. Give the proof that the DNA is the true genetic material.
5. How many of the 64 triplet permutations can be made from the four nucleotides A, U, G and C containing a) no uracils b) one or more uracils?
6. Let us assume that a single strand in a certain small section of the DNA, molecule has its bases in the following order: adenine, thymine, adenine, guanine, cytosine, guanine. In what order would the bases occur in the partner helix?

Chapter 22

Protein Synthesis

Genes are highly specific in function. Certain genes produce very specific proteins (enzymes and specific antigens). The enzymes are very specific in their action. The enzymes are the basic unit of the biochemists and the genes are the basic unit of genetist. Genes are specific in action and enzymes are also specific. Hence each gene is able to bring its action through a specific enzyme. This hypothesis is called "onegene-one enzyme hypothesis".

Genetic code:

The key to unlock the mistery of life lies in the fundamental functional unit of enzyme on one hand and the gene on the other hand. The building blocks from which the proteins are composed of amino acids. There are 20 different essential amino acids. All amino acids have common structure in the form of 'C' atom called the carbon to which is attached a carboxyl group and an amino group, a hydrogen atom and a side chain.

The union of two amino acid residues form a dipeptide and three tripetides and if many amino acid residues unite, they give polypeptide. Natural proteins are made up of a single polypeptide or

The Genetic Code

Singlet Code

A
G
C
U

Doublet Code

AA	AG	AC	AU
GA	GG	GC	GU
CA	CG	CC	CU
UA	UG	UC	UU

Sequence of two letters
— 16 words

Triplet Code

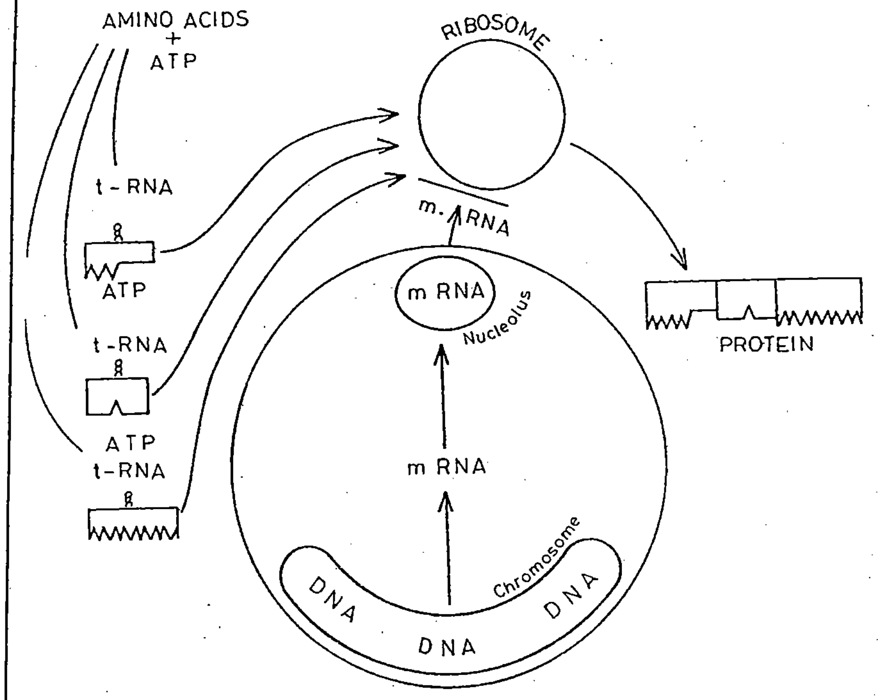
AAA	AAG	AAC	AAU
AGA	AGG	AGC	AGU
ACA	ACG	ACC	ACU
AUA	AUG	AUC	AUU
GAA	GAG	GAC	GAU
GGA	GGG	GGC	GGU
GCA	GCG	GCC	GCU
GUA	GUG	GUC	GUU
CAA	CAG	CAC	CAU
CGA	CGG	CGC	CGU
CCA	CCG	CCC	CCU
CUA	CUG	CUC	CUU
UAA	UAG	UAC	UAU
UGA	UGG	UGC	UGU
UCA	UCG	UCC	UCU
UUA	UUC	UUC	UUU

Sequence of three letters
- 64 words

THE GENETIC CODE

UUC UUC	Phenylalanine	UAU UAC	Tyrosine
UUA UUG CUU CUC CUA CUG	Leucine	UAA UUG	Stop
		CAU CAC	Histidine
		CAA CAC	Glutamine
AUU AUC AUA	Isoleucine	AAU AAG	Asparagine
AUG	Methoionine	AAA AAG	Lysine
GUU GUC	Valine	GAU GAC	Aspartic acid
GUA GUG		GAA GAG	Glumatic acid
UCU UCC UCA UCG	Serine	UGU UGC	Cysteine
		UGA UGG	Stop Eryptophan
CCU CCC CCA CCG	Proline	CGU CGC CGA CGG	Arginine
ACU ACC ACG ACG	Threonine	AGU AGC	Serine
		AGA AGG	Arginine
GCU GCC GCA GCG	Alanine	GGU GGC GGA GGG	Glycine

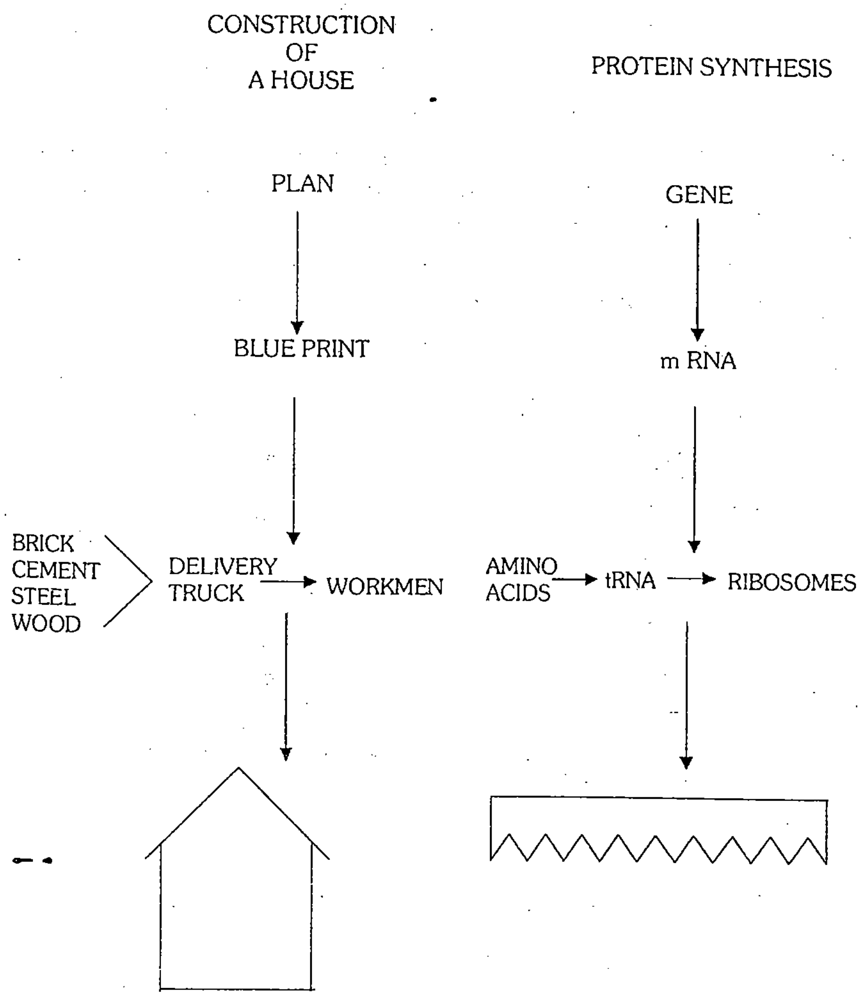
The triplets given are actually mRNA codons and are described in terms of RNA bases, namely
Uracil (U), Cytosine (C) Adenine (A) and Guanine (G)


AMINO ACIDS
+
ATP
RIBOSOME
t - RNA
ATP
t -RNA
ATP
t-RNA
m. RNA
m RNA
Nucleolus
m RNA
Chromosome
DNA
DNA
DNA
PROTEIN

Protein Synthesis

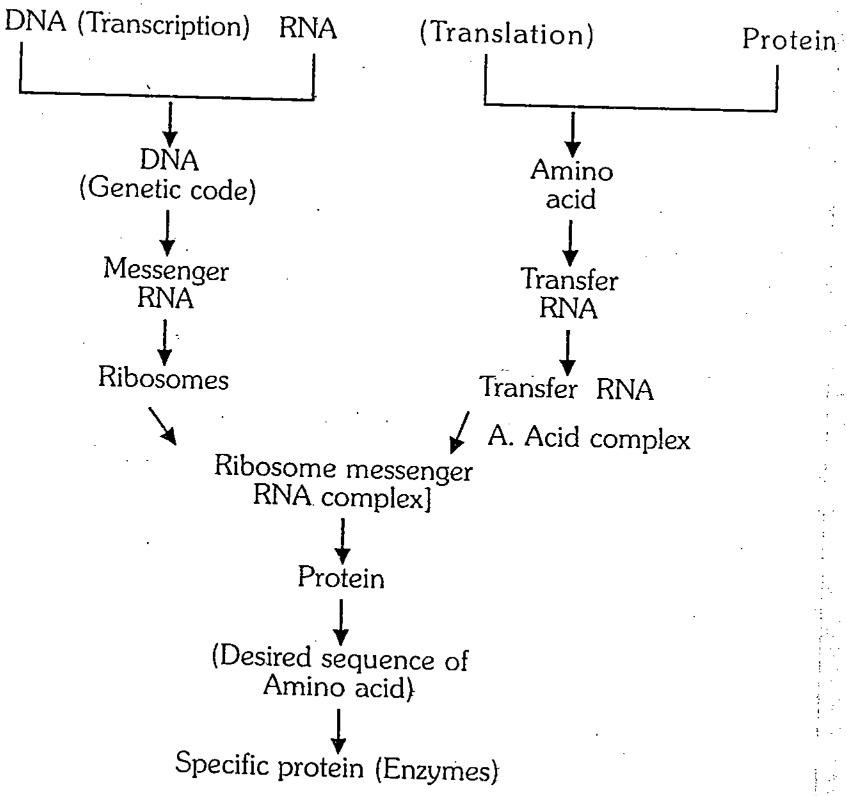
two or more polypeptides connected together by cross linkage between the side chains of certain of their amino acids. The specific protein (enzyme) is governed by the sequence of the amino acids residues in the polypeptide chain or chains of which they are composed of. Since there are 20 amino acids the number of different sequences of amino acid residues which is fixed constant for each protein is termed the primary structure of the protein. The coiling of the polypeptide chain is termed as the secondary structure of the protein. The shape and specificity of a protein molecule is based on the sequence of amino acids in the polypeptide chain. The ge-

PROTEIN SYNTHESIS


CONSTRUCTION
OF
A HOUSE
PROTEIN SYNTHESIS
PLAN
GENE
BLUE PRINT
m RNA
BRICK
CEMENT
STEEL
WOOD
DELIVERY
TRUCK
WORKMEN
AMINO
ACIDS
tRNA
RIBOSOMES

netic information specifying this sequence is carried by a region of chromosome (the gene) namely DNA in the form of chemical code. This chemical code is based on the sequence of four nucleotide bases of DNA. It is the code of letters (A, G, T, C) various combinations of this spells the word indicating each of the amino acid. This chemical code is called the genetic code. As per the genetic code the sequence of bases along the whole region of DNA lays down the sequence of amino acids in protein.

A sequence of 2 letters taken at a time can give a combination of only 16 words at a time. But there are minimum of 20 amino acids. Therefore a sequence of 3 letters which gives a combination of 64 words is sufficient to account the 20 amino acids and more. This sequence of 3 nucleotides determines a single amino acid and hence it is called "Triplet code". The Amino acids differ from a single code.



The genetic information (genetic code) for protein synthesis is carried from the genes (DNA) by the messenger RNA. The messenger RNA picks up its message in the form of a base sequence complimentary to that of DNA by being synthesised along one of the DNA strands in the presence of an enzyme the RNA polymerase. The messenger RNA when separates from its DNA primer and goes to a Ribosome to which it becomes reversibly attached.

The amino acids form complexes with adenylic acid in the presence of the ATP and specific activating enzymes. Each activated amino acid then unites with a specific transfer RNA molecule and is carried to Ribosome messenger RNA complex. It is assumed that some part of each transfer RNA molecule is characterised by a sequence of base complimentary to sequence on the messenger RNA which code for the particular amino acid which it carries.

Thus the transfer RNA molecules, each bearing its amino acid become arranged on template in the sequence originally prescribed by the DNA of the gene. Finally the amino acids are released from union with transferase and become incorporated into a polypeptide chain.

Point mutation:

A triplet code presumedly codes a single amino acid. If this triplet code were changed by substitution of different ways the code would be altered. Assume that the triplete code A~~,~~ C, B codes an amino acid X, a change of one base in this triplet can give A C D. Since ACD is a nonsense code, an amino acid would be omitted and either the formation of the protein would be stopped or the protein would be formed minus one amino acid. In either event such a change would be detected as a mutation at enzyme level. If the code was to change ACA then a sense triplet code is formed and an amino acid Y would be incorporated in the protein instead of the amino acid 'X' and this would have an altered protein (enzyme). Again, such a base substitution would appear as a mutation. Base substitution can have two general consequences.

1) ACB as the normal code.

2) ACD as the nonsense code which do not include any amino acid and

a missense code (ACA) which codes for different amino acid (a mutation).

Types of Genes:

Studies on the control of genetic expression had lead to the recognition of the two category of genes:

1) Structural genes and
2) Regulator genes.

A structural gene carries the genetic information and determines the structure and specifies function of a protein.

A regulator gene determines the production of cytoplasmic substance which controls the activity of specific structural genes (or) co-ordinated group of structural genes.

The regulator genes are concerned not only with enzyme systems but also with co-ordinated enzyme repression. Co-ordinated enzyme repression is also called as "Repression' which refers to the shutting off all enzymes of a sequence by its end products. The end products (metabolites) are resulting during the biosynthetic pathways. The regression of a particular pathway can be released as a result of a single mutation so that the production of the enzyme is no longer restricted by the presence of the end products.

Repressor genes are genes which bring about repression. Repressor genes must act by combining with some element called an operator. The operator is necessary for the transcription into protein of the genetic information contained in the group of genes.

Operator genes (or) operon:

This is a single operator that switches on a sequence of gene. The operator and structural genes control a compromise of an integrated unit at both the physical and functional level. The sperm is a genetic unit of the co-ordinated transcription. The genetic message is transmitted from the operon to the ribosomes through the messenger RNA.

Professor Harigovind Khorana - a fellow of the chemical society was

among American scientists who were awarded the 1968 Nobel prize. Khorana shared the award with Prof. Robert Holley and Dr. Marshall W. Nirenberg, all honoured for their independent contribution to the unrevelling of genetic code.

Professor Khorana working at the University of Wisconsin used Nirenberg's technique to elucidate further code words. He also synthesised a DNA of known sequence by linking two DNA bases and polymerizing to give a DNA chain containing an alternate sequence of the bases. This method allowed Khorana to prove conclusively that each base triplate was a code word in itself and was not overlapping, with preceding or succeeding triplets. Khorana's work represent a major step in the possibility at synthesising genetic material.

EXERCISE

1. Describe in detail on Genetic code.
2. Define the following:
    a) Triplet code
    b) Transcription
    c) Translation
    d) Ribosome RNA
    e) Transfer RNA
    f) Nonsense code
3. What are the different types of genes and describe on them?
4. Give an account on the research work done by Professor Khorana.
5. Write in detail on Protein synthesis.

Extranuclear Genetics

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Chapter 23

Cytoplasmic Inheritance

maternal inheritance

Genes are the fundamental units of inheritance. However, one should remember that the actual protoplasmic link between an individual and its two parents is more than a complement of genes or chromosomes or even a pair of nuclei, because the egg contributes a mass of cytoplasm, larger than either gamete nucleus. Quite a number of observations and experiments in the recent years suggest that some genetic variability is the result of self duplicating, mutable units located in the cytoplasm. These units are apparently transmitted only by means of cytoplasm and called as cytoplasmic inheritance. Since only the egg contributes cytoplasm this inheritance is also referred as maternal inheritance.

One type of cytoplasmic inheritance in plants is plastid inheritance. Plastids are cytoplasmic bodies occurring in certain cells of plants. Normal plants contain chlorophyl, the green colouring matter which is involved in the process of photosynthesis. Plastids are self perpetuating bodies within the cytoplasm which develop and function in conjugation with nuclear genes. The cytoplasm of the ovule which is incorporated into a seed carries primordia of the plastids which in certain respects determine what type of plastid develop in plant growing from the seed. The sperm from the pollen grain which carries very little cytoplasm does not influence the plastids.

In certain strains of Four O'clock plants, colourless plastids are found as well as green, but both types occur in the same plant and even in the same leaf. Such leaves are variegated i.e. irregularly blotched green and white. In some cases the spots may form small areas of the leaves, but in other cases entire leaves or branches may be affected. Seeds taken from a branch which are entirely green will produce plants which are white, but the white plants die because of lack of chlorophyl. Seeds taken from navigated branches produce green, white and navigated plants.

Pistel X Pollen Green or white
Green | Green or white
Green only

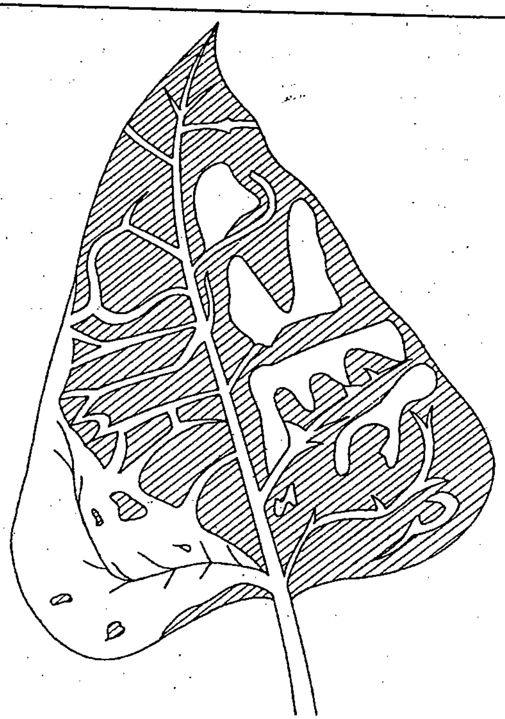
Pistel X Pollen
White | Green or white
White only

Pistel X Pollen
Navigated Green or white
Green, white and
navigated

In the above crossings, the offsprings resemble only the colour of the pistel (Mother). Since plastids are present in cytoplasm and ordinarily no cytoplasm is brought in by the sperm (pollen), this is a true case of cytoplasmic inheritance.

Example in Drosophila:

One strain of Drosophila was found to be more sensitive to carbondioxide. This particular strain of Drosophila is affected by much smaller concentration than those require to anaesthetize normal strains and may be killed by stronger concentrations. This strain is known as CO_2 sensitive strain. The cross between normal and CO_2 sensitive strain is shown below:



A Leaf of Variegated Four O'Clock Plant

CO_2 sensitive strain X Normal strain
Female | Male
Highly sensitive to CO_2

CO_2 sensitive strain X Normal strain
Male | Female
Normal

The results clearly indicate that the transmitting factor for CO_2 sensitization is located in the cytoplasm only. The self perpetuating unit is known as sigma. This is a typical example of cytoplasmic inheritance.

Example in Paramecium:

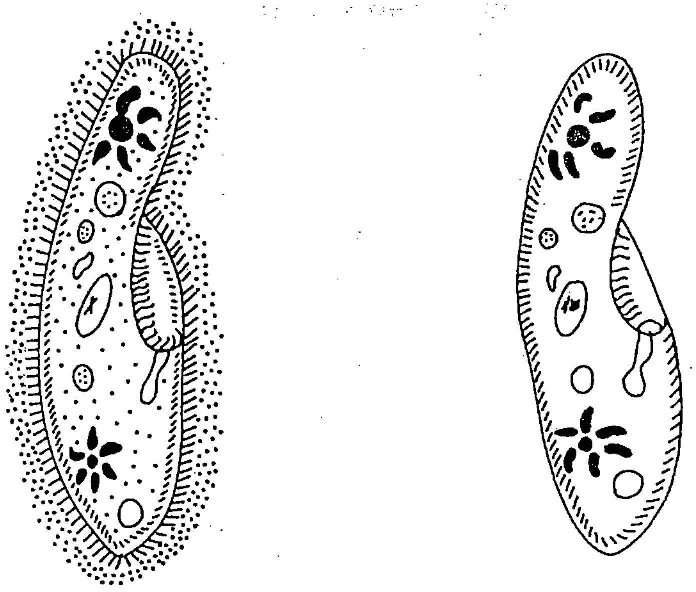
T.M. Sonneborn had conducted experiments on Paramecium, a one celled protozoan which is in abundance in ponds. It reproduces asexually by binary fission and sexually by conjugation. In conjugation two organisms come together and form a temporary bridge of cytoplasm between them and exchange nuclear material over this bridge.

When different strains of Paramacium amelia were mixed in a culture dish to allow conjugation to occur, it was found all the members of one strain died and the others survived. The strain that survives is known as killer strain. Sensitives produced sensitives and killers produce killers. The substance liberated by the killer strain is called 'Parameim'. The substance in cytoplasm which generates paramecium is designated as kappa. Kappa particles are seen only in the cytoplasm of killer strain. No kappa particles are present in the cytoplasm of sensitives. Kappa can be maintained in an individual and its progeny only when the dominant gene K is present in the nucleus.

KK } Killers kk - sensitives
Kk }

However, the mere presence of the gene K in the nucleus does not insure the presence of kappa. The kappa articles are self duplicating and must come from pre-existing kappa only. A killer strain must contain both the gene K and a suitable concentration of kappa. The concentration of kappa particles to make an individual killer is about 400 to 1,000 particles. In one type of sexual reproduction, two individuals of opposite mating type come together and exchange one of the two micronuclei which each one possesses. The other type of sex reproduction involves only one parent known as Autogamy. Because of these different modes of reproduction it is possible to manipulate paramecim in such a way to combine any cytoplasm with any desired gene. Thus it is possible to replace gene K by its allele k in a killer animal. When this is done, the paramacium gradually loses its kappa and becomes sensitive.

Kappa Particles in Paramecium

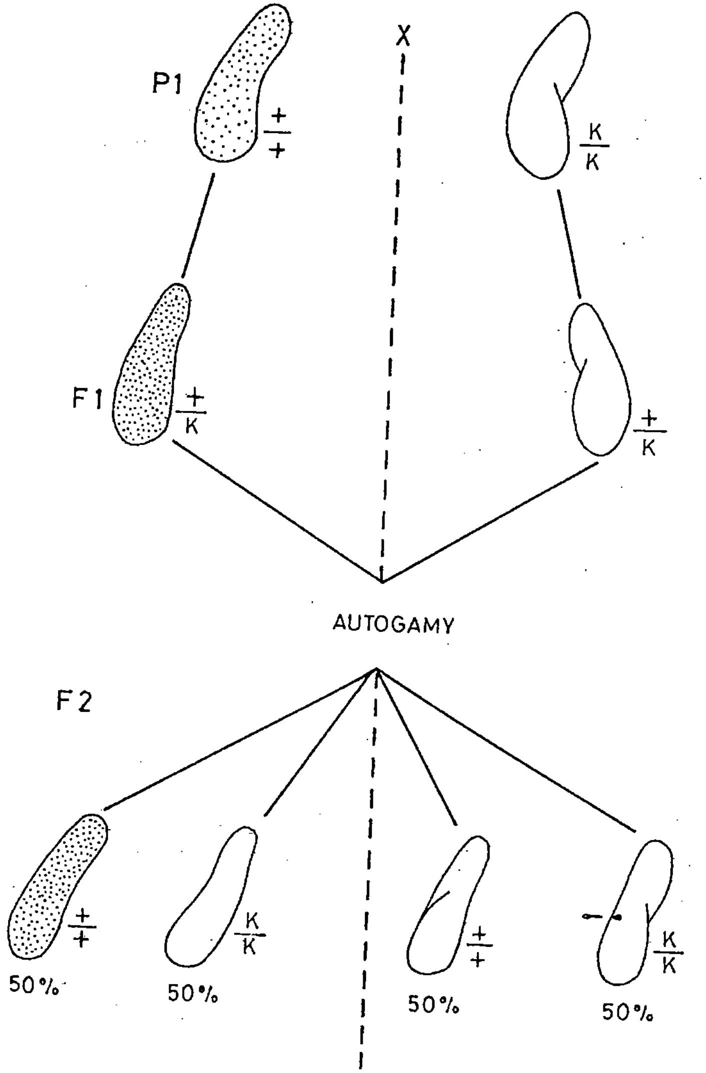


Killer with Kappa particles inside and paramecium in the liquid medium outside the organism Gene - K is present in the nucleus

Sensitive paramecium with no Kappa particles, no paramecium and genes — kk in the nucleus

If a killer KK conjugates with sensitive kk, the conjugates are kk and should be killers, but if the conjugation is brief, and no cytoplasm is exchanged, then a killer clone and a sensitive clone are produced. A more prolonged conjugation results in exchange of cytoplasm and all the descendent clones are killers since they have kappa particles. Thus the Kk individuals are all killers. The kappa particles are clearly transmitted through the cytoplasm only.

Inheritance of Killer Character in Paramecium



Killer Gene K is transmitted but not the iller Cytoplasm.

EXERCISE

1. Describe on cytoplasmic inheritance.
2. Why the cytoplasmic inheritance is also termed as maternal inheritance?
3. Give an account on Plastid inheritance.
4. Describe in detail on the cytoplasmic inheritance in Paramacium.
5. Continued use of certain drugs can lead to addiction. Addicted mothers by non-addicted fathers give birth to babies which are addicted. The children of addicted fathers by non-addicted mothers are normal. The difference in reciprocal crosses is not due to extranuclear genetic elements. Suggest a mechanism to account for the results.
6. Many strains of chlamydomonas are sensitive to streptomycin. A strain is found which requires streptomycin in the culture medium for its survival. How could it be determined whether streptomycin-dependence is due to a chromosomal gene or to a cytoplasmic element?
7. A Yeast culture when grown on medium containing acriflavin, produces numerous minute cells which grow very slowly. How could it be determined whether the slow growth was due to a cytoplasmic factor or to a nuclear gene?
8. Some Drosophila flies are known to be very sensitive to Co_2 gas, rapidly becoming anaesthesised under its influence. This trait shows maternal inheritance. Predict the results of a cross between a) sensitive female x resistant male b) Sensitive male x resistant female.
9. When a killer strain of paramecia conjugates without cytoplasmic transfer with a sensitive strain, half of the exconjugants are sensitives and half are killers. Autogamy of the killer exconjugants produce only killers, autogamy of the sensitive exconjugants produce only sensitives. What are the genotypes of
   a) the two original strains?
   b) the exconjugants?
   c) the autogamous products?

Population Genetics

Chapter 24

Population Genetics

A population is defined in genetic sense as a breeding group of individuals. It may be a breed or species or some group of individuals inhabiting a locality. A study on the genetics of such a population is referred as Population Genetics. It is concerned with the inheritance of those differences between individuals in quantitative characters and hence it is also known as Quantitative Genetics.

The Classical Mendelian traits have been qualitative in nature, which are easily classified into distinct phenotypic categories. These qualitative traits are under the control of only one or very few genes with little or no environmental modifications.

In contrast to this, quantitative traits fail to fit into separate phenotypic classes, but instead form a spectrum of phenotypes. Economically important traits such as body weight, weight gain, milk production, wool yield, egg yield etc. are quantitative traits with continuous variability.

The basic differences between Qualitative and Quantitative traits are:

Qualitative traits	**Quantitative traits**
1. Characters of kind	Characters of degree
2. Discontinuous variation-discrete phenotypic classes	Continuous variation-Phenotypic measurements form a spectrum
3. Single gene effects can be detected	Polygenetic control effects of single genes too slight to be detected
4. Concerned with individual matings and their progeny	Concerned with a population of organisms, consisting of all possible kinds of matings
5. Analysed by making counts and ratios	Analysed by mean and standard deviation

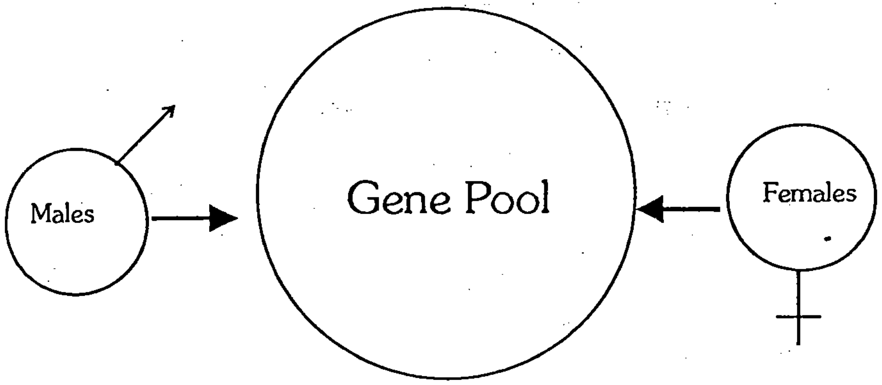
An understanding of quantitative traits inheritance is of fundamental significance in the study of evolution and in the application of genetics to animal breeding.

In quantitative traits, the individual genes whether few or many can not be identified by their segregation. The Mendelian ratios are not displayed and hence Mendelian analysis can not be applied. But the quantitative traits depend on genes subject to the same laws of transmission and have the same general properties as that of qualitative traits. Hence quantitative genetics or population genetics is an extension of Mendelian Genetics resting on Mendelian principles as its foundation.

The population or quantitative genetics are different in two aspects.

1) Since ratios can not be observed and single progenies are uninformative, the study must be extended to large group of individuals having many progenies i.e. population.

2) The study requires measurement and not just classification of the individuals.



Contribution of Males and Females to Gene Pool

The first aspect indicates that the study of inheritance of quantitative traits extended to population, hence it is referred as Population Genetics. As per the second aspect, the study require measurements of traits, and so it is also called as Biometrical Genetics.

PROPERTIES OF POPULATION

A population is defined as breeding group of individuals.

Genetics of a population is concerned not only with the genetic constitution of the individuals but also with the transmission of genes from generation to generation. In the transmission, the genotypes of the parents are broken down and a new set of genotype is constituted in the progeny from the genes transmitted in the gametes. The genes are having continuity but the genotypes are not having that continuity.

Genetic properties of population:

Males and females contribute their genes to a gene pool.

Males ⟶ Gene or Gamete pool ⟵ Female

A gene pool is the total genes possessed by males and females of a population. To describe the genetic constitution of a population, we may have to specify the genotype and the genes carried by the population.

Genotypic frequency is the proportion or the percentage of genotypes in the population.

Example: Let us consider the locus controlling coat colour in short horn cattle. Where the Red (R) and white (r) alleles are involved. With the two alleles at a locus, there could be 3 possible genotypes namely RR, Rr and rr. Red colour would be of genotype RR, roan— Rr and white — rr.

Let us assume that the total herd strength is 100 with

RR - 30, Rr - 60 and rr - 10.

The genotypic frequencies of the population could be 30, 60, 10

In percentage 30%, 60%, 10%

When expressed in frequencies they add up to unit 0.3 + 0.6 + 0.1 = 1.

When expressed in percentage they add upto 100.

Gene frequency:

The genetics of a population is not only concerned with the distribution of genotypes but also with the transmission of genes from one generation to the next.

Gene frequency is defined as the number of genes present in the population. It is expressed as proportion or percentage.

The frequency of all alleles carried by any locus add upto unity or 100 i.e. the frequency of R gene and r gene add upto unit or 100% in the herd.

The gene frequency can be estimated from the knowledge of genotypic frequencies.

Let us estimate the gene frequency in short horn herd.

Phenotype	Red	Roan	White	Total
Genotype	RR	Rr	rr	
No.of individuals in the genotype	30	60	10	100
No.of Genes in (R)	60	60	-	120
(r)	-	60	20	80
Total no. of genes				200

Each individual has 2 genes. Therefore a total of 100 x 2 = 200 genes are carried in the population of 100 individuals.

Each RR individual carry two R genes. Similarly each rr individual carry two r genes. Each Rr individual carry one R gene and one r gene. Thus there are 30x2+60x1=120 R and

10x2+60x1 = 80 r gene.

The frequency of R gene is 120/200=0.6 or 60% and r gene is 80/200=0.4 or 40%.

Let us assume the frequency of 'A' gene in a locus containing two alleles A and 'a'. The frequency of 'A' gene be p

The frequency of gene 'a' be q

	Gene		Genotypes		
	A	a	AA	Aa	aa
Frequencies	p	q	P	H	Q

$p + q = 1$

$P + H + Q = 1$

Genes in Parents

	A_p	a_q
A_p	p^2	pq
a_q	pq	q^2

$p^2+2pq+q^2= 1$

Estimation of Gene frequency from Genotypic frequency:

Let us assume that there are two alleles A, a at a locus. Further let us assume that there are N diploid individuals of which P are dominant (AA), H are heterozygous (Aa) and Q are recessive (aa), where P+H+Q=N. Since each individual carry two genes there are altogether 2N genes in the given population of N individuals. P number of individuals carry two A genes, H number of individuals carry one A gene and one a gene, and Q number of individuals carry two a genes.

The gene frequencies of the population are:

The gene frequency - A-gene = 1/2N (2P+H)

The gene frequency - a-gene = 1/2N (H+2Q)

Example: Find the gene frequency in a group of 40 individuals.

AA	Aa	aa
2	12	26

Frequency of A Gene = P =1/2x40(2x2+12) = 0.20

Frequency of a Gene = q = 1/2x40(12+26x2) = 0.80

Generally, the frequency of genotypes may be given in percentage or proportions instead of in numbers especially when the group is large. Then P + H + Q = 1. The gene frequencies p and q can then be estimated using the following equation:

p = 1/2 (2P+H) = P+1/2H

q = 1/2 (H+2Q) = Q+1/2H

Example: The population given in the above example can be represented in terms of proportions as 0.05, 0.30 and 0.65. The gene frequencies can be estimated as follows:

P = 0.05 + 1/2 (0.30) = 0.20

q = 1/2 (0.30) + 0.65 = 0.80

To understand the transmission of genes from one generation to the next in a population, let us take the example of coat colour in short horn.

Phenotype	Red	Roan	White	Total
Genotype	RR	Rr	rr	
No.of individuals	30	10	60	100
No. of Genes R	60	10	-	70
r	-	10	120	130

Let the frequency of gene R as p and the frequency of gene r as q. The Zygotic frequencies will be formed by the combination of gametes as shown below:

Parents

O	R_p	r_q
R_p	RR p^2	Rr pq
r_q	Rr pq	rr q^2

The zygotic frequencies of RR, Rr and rr zygotes will be p^2, 2 pq and q^2 respectively. In general the zygotic array produced under random union of gametes is the square of gametic array.

$$(p+q)^2 = p^2 + 2pq + q^2$$

The frequency of R gene = p = $p^2+1/2(2pq) = p^2+pq$

The frequency of r gene = q = $q^2+1/2(2pq) = q^2+pq$.

EXERCISE

1. Define the following:
    - a) Population
    - b) Quantitative traits
    - c) Biometrical genetics
    - d) Population genetics
    - e) Genotypic frequency
    - f) Gene frequency
2. In a short horn herd there were 72 red, 100 roan and 28 white individuals. What is the frequency of genotypes and genes in the population?
3. A population of cattle have 36% homozygous polled. What will be the gene and genotype frequencies of polled and horned condition?
4. Estimate the gene and genotypic frequencies of the following population of short horn cattle:

    Red cows - 49   White cows - 9   Roan cows - 42
5. Find the gene frequency in a group of 40 individuals.

    AA-2   Aa-12   aa-26
6. Find the gene frequencies of the following proportions.

    AA-0.05   Aa-0.30   aa-0.65

Chapter 25

Hardy-Weinberg Equilibrium

AGENCIES CAUSING CHANGE IN GENETIC PROPERTIES OF POPULATION

The genetic properties of population are influenced by number of agencies in the process of transmission of genes from one generation to the next. Few agencies which cause changes are listed below.

1) Population size:

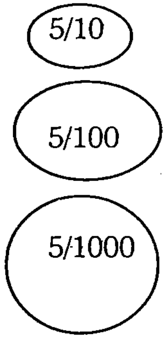
The genes passed from one generation to the next are from sample of genes in the parent generation. Hence the gene frequencies are subjected to sampling variation. If it is a large population, sampling variation will be so small as to be negligible. Large population is the one in which the number of adult individuals will be in hundreds rather than in tens.

For example, five horned cows in different population sizes shall indicate varying proportions.

5/10 = 50%

5/100 = 5%

5/1000 = 0.5%



The gene and genotypic frequencies depend on the population size. Smaller the size of population greater the sampling variations.

2) Differences in fertility and viability:

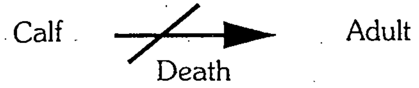
The individuals differ in fertility. Some individuals produce more number of offsprings, some produce less number and some do not produce at all. For example, the cows, A, B, C, and D may be different in producing the number of calves.

Cows

A	B	C	D
5 calves	2 calves	1 calf	0 No calving

If an individual is not having any progeny, that means the genes in him are not transmitted to the next generation. It is referred as Genetic death. In the above example animal D has not given birth, and thus genes from D have not been transmitted to the next generation.

The viability of the individual may also affect the gene and genotypic proportions of the population. The progeny of one individual if not viable,then transmission of genes to the next generation is interrupted.



Occurrence of death in a calf results in non-transmission of its genes. In this way, the gene frequency may be changed in the transmission.

3) Migration:

Migration means movement of individuals. It may be introduction (immigration) of individuals into the population or elimination (emigration) of individuals from the population. The gene and genotypic frequencies may be changed by both immigration and or emigration.

4) Mutation:

Sudden change in the structure of gene is mutation. It may be from wild type to mutant type or vice versa. Both forward and reverse mutations may alter the gene frequencies of population.

5) Selection:

Choosing the parents of next generation is selection. It may be natural or artificial selection. Any type of selection may alter the gene frequencies of the population. Selection favouring or unfavouring certain genes may increase or decrease the frequency of genes.

6) Mating systems:

The genotypes in the progeny are determined by the union of gametes of the parents and the union of gamates is influenced by the mating of the parents. The genotypic frequencies of progeny generation are influenced by the genotypes of parents. The non-random mating or selective mating may alter the frequencies whereas random mating or Panmixia may keep the frequencies constant. The non-random mating includes assortative and disassortative matings.

HARDY-WEINBERG LAW

Hardy - a British scientist, and Weinberg - a German scientist proposed this principle independently in 1908. This law states, "In a large random mating population, in the absence of migration, mutation and selection the gene and genotypic frequencies remain constant from generation to generation".

A population with constant gene and genotypic frequencies is said to be in Hardy-Weinberg equilibrium.

Proof of Hardy-Winberg law with constancy of Gene frequency:

The proof involves four steps.

1. Production of Gametes:

Let the genotypes of the parents be AA, Aa and aa and let the frequencies of 'A' gene be 'p' and 'a' gene be 'q'. The genotpic frequencies shall be as follows:

Genotype	AA	Aa	aa
Frequency	P	H	Q

The individual AA shall produce only one type of gamete carrying 'A' gene. Similarly aa individuals shall produce only one type of gamete with a gene. Two types of gametes will be produced by Aa individuals, 'A' and 'a' gametes. Hence the gene frequency of A = P+1/2H. Thus the gene frequency in the whole gametic output is the same as in the parents.

2) Formation of Zygotes:

Random mating between individuals is the random union of their gametes. The genotypic frequencies among the zygotes are then products of the frequencies of gametic types that unite to produce them. The genotypic frequencies among the progeny produced by random mating can be determined by multiplying the frequencies of gametic types produced ~~by~~ each sex of the parents.

A a
2P a

Female parent		Male parent A_p	Gametes and their frequencies a_q
Gametes and their frequencies	A_p	p^2	pq
	a_q	pq	q^2

The frequencies of zygotes

AA	Aa	aa
p^2	$2pq$	q^2

3) Viability of Zygotes to adults:

The frequencies will be observable only when the zygotes survive and grow as adult. Then only the genotypes can be classified.

4) Gene frequency in Progeny:

The genotypic frequencies of progenies are

AA	Aa	aa
p^2	$2pq$	q^2

The gene frequency of A in the progeny:

$$p = p + \frac{1}{2}H$$

$$= p^2 + \frac{1}{2}(2pq)$$

$$= p^2 + pq$$

$$= p(p+q)$$

(since $p+q = 1$)

$$= p\times1 = p$$

The gene frequency of A in the progeny generation is = p

Similarly gene frequency of a in the progencies

$$a \quad = q = Q + \frac{1}{2}H$$

$$= q^2 + \frac{1}{2}(2pq)$$

$$= q^2 + pq$$

$$= q(q+p)$$

Since $q + p = 1$

$$= q\times1$$

$$= q$$

The frequency of A is p and of a is q as in the parents. This proves the constancy of the gene frequency from one generation to the next.

Proof of Hardy-Weinberg law with constancy of Genotypic frequency:

Let us consider a locus with two alleles, A and a, and let the frequencies of genes and genotypes in the parents be as follows:

	Genes		Genotypes		
	A	a	AA	Aa	aa
Frequencies	p	q	P	H	Q
			p^2	2pq	q^2

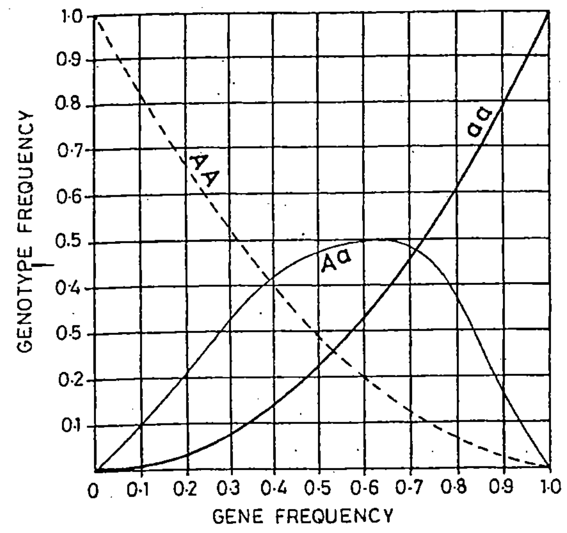
Genotype frequency of Male parent

			AA	Aa	aa
Genotype frequency of female parent	AA	P	P	H	Q
			p^2	PH	PQ
	Aa	H	PH	H^2	HQ
	aa	Q	PQ	HQ	Q^2

—There are altogether nine types of mating and their frequencies when mating is at random are found by multiplying the frequencies. Some of the types of mating are equivalent, the number of types reduces to six. By summing up the frequencies of equivalent types, the frequencies of mating types are obtained.

The genotypes of total offsprings are considered by adding the frequencies of the genotype of offspring produced by each type of mating. For example mating of type AA x AA produce only AA offspring. So of the total progeny a proportion p^2 are AA genotypes produced from this type of mating.

Mating		Genotype and frequency of progeny		
Type	Frequency	AA	Aa	aa
AA x AA	P^2	P^2	-	-
AA x Aa	2PH	PH	PH	-
AA x aa	2PQ	-	2PQ	-
Aa x Aa	H^2	$(1/4)H^2$	$(1/2)H^2$	$(1/4)H^2$
Aa x aa	2HQ	-	HQ	HQ
aa x aa	Q^2	-	-	Q^2
Total:		$(P^2+PH+1/4H^2)$	$(PH+2PQ+1/2H^2+HQ)$	$(1/4H^2+HQ+Q^2)$



Gene frequency of a

Relationship between genotype frequencies and gene frequencys for two alleles in a population under Hardy-Weinberg equilibrium

To find out the frequency of each genotype in the total progeny, the frequencies contributed by each type of mating are added.

$$= (P^2+PH+1/4H^2) \quad + \quad (PH+2PQ+1/2H^2+HQ) \quad + \quad (1/4H^2+HQ+Q^2)$$

$$= (P+1/2H) \quad + \quad 2(P+1/2H)(Q+1/2H) \quad + \quad (Q+1/2H)^2$$

$$= \quad p^2 \qquad\qquad 2pq \qquad\qquad q^2$$

After simplification, the genotypic frequencies of progenies AA, Aa and aa are p^2, 2pq and q^2 respectively which are the same genotypic frequencies of the parents. Thus the genotypic frequencies remain constant from generation to generation.

Properties of Hardy-Weinberg law:

1) A population of random mating in the absence of migration, mutation and selection will be under Hardy-Weinberg equilibrium.
2) The genotypic frequencies of the progeny depend only on the gene frequencies of the parents and not on the genotypic frequencies.
3) One generation of random mating will produce the population under Hardy-Weinberg equilibrium.
4) The frequency of heterozygotes will not be greater than 0.5 i.e. 50%.
5) When the gene frequency of an allele is low, the rare allele occurs predominantly in the heterozygotes and less in homozygotes.

Uses of Hardy-Weinberg law:

1) The gene frequency of heterozygotes of or carriers can be estimated.

For example, in human beings, albinos are recessive. Let the proportion of albinos be one in 10,000. The proportion of heterozygous individuals or carriers can be estimated.

The proportion of albinos $= \frac{1}{10,000}$

Let it be aa = its frequency be q^2

When $q^2 = 1/10,000$

$q = \sqrt{1/10,000} = 1/100 = 0.01$

Since p+q = 1

The frequency of A gene = p = 1-q

= 1-0.01

= 0.99

The proportion of heterozygous

individuals = 2pq

= 2 x 0.01 x 0.99

$= \frac{198}{10,000}$

2) The gene frequency of multiple alleles can be estimated. (Eg.) In a human population of 100 individuals, A, B, O and AB blood groups are distributed as follows:

Blood groups	A	B	O	AB	Total
No.of individuals	32	15	49	4	100

Let the frequency of genes A be p

B be q

O be r

Then p + q + r = 1

Blood groups	A		B		O	AB
Genotypes	AA	AO	BB	BO	OO	AB
Frequency	p^2	2pr	q^2	2qr	r^2	2pq

Blood group	O
Genotype	OO
Frequency	r^2

No. of individuals 49/100 or 0.49

$r^2 = 0.49$

$r = \sqrt{0.49} = 0.7$

Blood groups = B+O

Genotypic frequency $= q^2 + 2qr + r^2$

$= (q+r)^2$

$(q+r)^2 = B+O$

$q+r = \sqrt{B+O}$

$= \sqrt{0.15 + 0.49}$

$= \sqrt{0.64} = 0.8$

Since $p+q+r = 1$

$q+r = 1 - p$

$1-p = 0.8$

$p = 1 - 0.8 = 0.2$

Genotypic frequency of Blood groups

$A+O = P^2 + 2pq + r^2$

$A+O = (p+r)^2$

$\sqrt{A+O} = p+r$

$\sqrt{0.32 + 0.49} = p+r$

$\sqrt{0.81} = p+r$

Since $p + q + r = 1$

$p + r = 1-q$

$1-q = \sqrt{0.81}$

$= 0.9$

$q = 1 - 0.9 = 0.1$

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Hence the frequency of gene A = p = 0.2
B = q = 0.1
O = r = 0.7

3) Hardy-Weinberg law is useful to test a population whether it is in Hardy-Weinberg equilibrium or not.

Example: In a herd of cattle there are 72 red, 100 roan, and 28 white individuals. The population has to be tested for Hardy-Weinberg equilibrium.

Let the gene and genotypic frequencies be as given below:

Phenotype	Red	Roan	White
Genotype	RR	Rr	rr
No.of individuals	72	100	28
Frequency	P	H	O

Let the frequency of gene = R be p and the frequency of gene = r be q.

Frequency of R $= p = P + \frac{1}{2}H$

$= 72 + 1/2\ (100)$

$= 72 + 50 = 122$

$= 122/200 = 0.61$

Frequency of r $= 1-p$

$= 1 - 0.61$

$= 0.39$

Expected frequency of Genotypes

	RR	Rr	rr	
	p^2	$2pq$	q^2	
	(0.61) (0.61) x200	(2x0.61x0.39) x200	$(0.39)^2$ x200	Total
Expected Numbers	74	96	30	200
Observed Number	72	100	28	200

Chisquare Test is applied to find out the significance of the differences.

$$\text{Chisquare Test} + \frac{(\text{Observed - Expected})^2}{\text{Expected}}$$

$$
\begin{aligned}
X^2 &= \frac{(O\text{-}E)^2}{E} \\
&= \frac{(72\text{-}74)^2}{74} + \frac{(100\text{-}96)^2}{96} + \frac{(28\text{-}30)^2}{30} \\
&= \frac{(-2)^2}{74} + \frac{(4)^2}{96} + \frac{(-2)^2}{30} \\
&= \frac{4}{74} + \frac{16}{96} + \frac{4}{30} \\
&= 0.39
\end{aligned}
$$

Table value of X^2 test at 5% level is 3.84. To be significant, the calculated value should be greater than the table value. In the above example, the calculated value is lesser than the table value. Hence the given population is under Hardy-Weinberg equilibrium.

The conditions necessary for maintenance of Hardy-Weinberg equilibrium in a population:

When the gene and genotypic frequencies of a population remain constant from generation to generation, the population is said to be under Hardy-Weinberg equilibrium. There are certain conditions required to maintain the population under Hardy-Weinberg equilibrium.

1) Mating must be at random:

Random mating is that in bisexual organisms, any one individual of one sex is likely to mate with any other individual of the opposite sex. In other words, the frequency of mating is dictated by chance.

2) The population should be large:

Genes that pass on from one generation to the next are samples of the genes in the parent generation. Therefore the gene frequencies are subjected to sampling variation between successive generations and smaller

the number of parents, the greater will be the sampling variation. Hence in large populations, the sampling error is so small as to be negligible.

3) Migration, mutation and selection must be absent:

The gene frequency of a population will get altered by the immigrants carrying a different gene frequency, mutation and selection against a particular allele or genotype.

Sex linked genes and Hardy-Weinberg equilibrium:

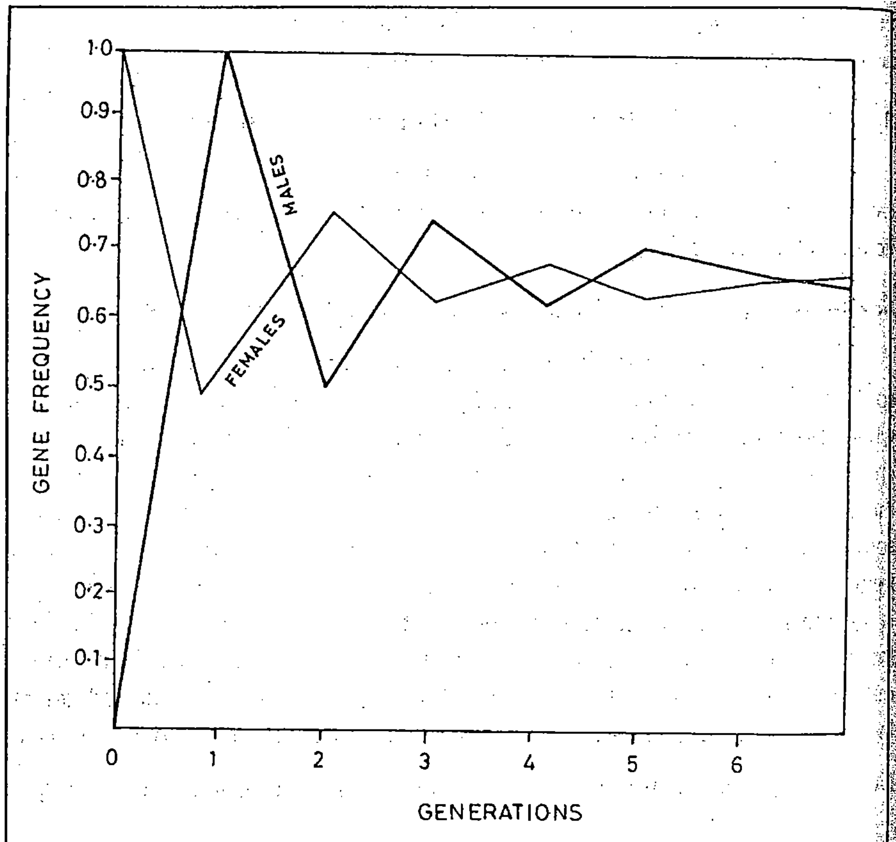
Sex linked genes are borne on x chromosome. In mammals and Drosophila, the females are homogametic i.e. they have two x chromosomes while the males are heterogametic having one x chromosome and one y chromosome. The males transmit the x chromosome to his daughters and the y chromosome to his sons. The female transmit a random x chromosome to her offspring regardless of their sex. The relationship between gene frequency and genotypic frequency in the homogametic sex i.e. in females is the same as that of autosomal genes, but in heterogametic sex (males) there are only two genotypes instead of three and each individual carries only one gene instead of two. Out of the total number of sex linked genes in a population, 2/3 of genes are carried by females and 1/3 genes by males.

Example: Consider two alleles A and a with the frequencies of p and q.

	Females			Males	
Genotype	AA	Aa	aa	AY	aY
Frequency	P	H	Q	R	S

Frequency of A in females (p_f) = P+1/2H

Frequency of A in males (p_m) = R



Sex Linked Gene Frequency under Hardy-Weinberg Equilibrium

Frequency of A in whole population

$$= 2/3 \text{ pf} + 1/3 \text{ pm}$$

$$= 1/3\ (2\text{pf} + \text{pm})$$

$$= \{1/3\ 2(P+1/2H)+R\}$$

$$= 1/3\ (2P+H+R)$$

Because of the asymmetrical chromosomal complement of males and females in the case of sex linked genes, the equilibrium state is not established in just one generation of random mating as was found with the autosomal genes. Gene frequency among males and females are different and hence the population is not in equilibrium. The distribution of gene frequencies between the two sexes oscillates as the population approaches equilibrium. Males get their sex linked genes only from their mothers. Therefore pm = pf in the previous generation. Females get their sex linked genes equally from both parents.

Therefore pf = pm+pf/2 = mean of the pm and pf in the previous generation.

The distribution of genes between male and female sexes oscillate, but the difference is halved in successive generations and the population rapidly approaches equilibrium in which the frequencies in the two sexes are equal.

Example: Assume the sex linked gene in females pf as 1 and pm as O, in the base generation. Then the pf and pm in the successive generations will oscillate and reach equilibrium in the sixth generation.

Generation	**pf**	**pm**
0	1	0
1	(1+0)/2=0.5	1
2	(1+0.5)/2 = 0.75	0.5
3	(0.75+0.5)/2=0.625	0.75
4	(0.625+0.75)/2=0.6875	0.625
5	(0.6875+0.625)/2=0.65625	0.6875
6	(0.6875+0.65625)/2=0.67	0.656
7	(0.67+0.656)/2 = 0.66	0.67

Multiple genes and Hardy-Weinberg equilibrium:

With multiple genes, equilibrium is not established in one generation of random mating but is approached very rapidly in subsequent generations. If there is linkage, the approach to equilibrium will be slower than if they are independent. Higher the intensity of linkage, slower the rate of approach to equilibrium. Disequilibrium with respect to two or more loci is called gametic phase disequilibrium or linkage disequilibrium.

Disequilibrium is due to

a) Intermixture of populations with different gene frequencies.

b) Chance in small populations.

c) Selection favouring one combination of alleles over another.

EXERCISE

1. What are the factors or agencies influencing the properties of a popualtion?
2. Define Hardy-Weinberg law. Prove the law with the constancy of gene frequency and genotypic frequency.
3. Give an account on the properties of Hardy-Weinberg law and its uses.
4. When the popualtion is said to be under Hardy-Weinberg equilibrium, what are the conditions necessary for its maintenance?
5. Describe on the frequency of sex linked genes under Hardy-Weinberg equilibrium.
6. How and to what extent do mutation, selection and migration affect the distribution of a relatively rare allele in a random population?
7. In Drosophila 'ee' gives ebony colour and EE, Ee gives grey colour. In a population with Hardy-Weinberg equilibrium 99 out of 100 flies are wild type. Calculate the frequency of ebony gene and the proportion of heterozygote in the population.
8. In Hereford cattle dwarfism is due to a recessive gene 'd'. Analysis of a herd book showed that out of every 100 calves born one is a dwarf. What is the frequency of genotypes DD, Dd and dd?
9. In a country the frequency of albinos is 1/10,000. What is the frequency of carriers and homozygous normal individuals in the population assuming random mating?
10. In a population gene pool, the alleles A and a are at initial frequencies p and q respectively. Prove that the gene frequencies and the zygotic frequencies do not change from generation to generation as long as the Hardy-Weinberg conditions are maintained.
11. Prove the Hardy-Weinberg law by finding the frequencies of all possible kinds of matings and from these generating the frequencies of

genotypes among the progeny using the symbols shown below.

	Alleles		Genotypes		
	A	a	AA	Aa	aa
Frequency	p	q	p^2	2pq	q^2

12. A large population has 0.04, 0.32 and 0.64 frequency of genotypes. Prove the gene and genotype frequency in the offspring remain the same as that of parent.

13. In a cattle herd, there were 72 Red (RR), 100 roan (Rr) and 28 white (rr) individuals. Test whether the population is in Hardy-Weinberg equilibrium.

14. A random sample of Arab population typed for MN blood groups have the following results:

Blood group	M	MN	N	Total
Frequency	119	76	13	208

Does this show that the population is in Hardy-Weinberg equilibrium?

15. In a human population of 100 individuals the A,B,O and AB blood groups are distributed as follows:

Blood groups	A	B	O	AB
No.of individuals	32	15	49	4

Calculate the gene frequencies of A,B and O genes. Test whether this population is under Hardy-Weinberg equilibrium.

16. Baldness is governed by a sex-influenced trait which is dominant in men and recessive in women. In a sample of 10,000 men, 7,225 were found to be non-bald. In a sample of women of equivalent size, how many non-bald women are expected?

17. The presence of horns in some breeds of sheep is governed by a sex-influenced gene which is dominant in males and recessive in females. If a sample of 300 female sheep is found to contain 75 horned individuals, (a) what percentage of the females is expected to be heterozygous, (b) what percentage of the males is expected to be horned?

18. In a population, the frequency of sex linked genes $q_f=0.6$ and $qm=0.4$, Calculate p. Find p_f and p_m for the first two genetations.

19. If $q_f=0.5$ and $q_m=0.5$, Find out p_f and p_m for the first and second generation.

20. In a population with sex linked recessive trait, proportion among females is 19.36% What % of males would you expect to show in this trait?

21. What is the proportion of colour blind women in a population having 8% of colour blind men?

22. A genetic disease of man called haemophilia (excessive bleeding) is governed by a sex linkd recessive gene which constitutes 1% of the gametes in the gene pool of a certain population.

    (a) What is the expected frequency of haemophilia among men of this population? What is the expected frequency of haemophilia among women?

23. Colour blindness in man is due to a sex linked recessive gene. A survey of 500 men from a local population revealed that 20 were colour blind? (a) What is the gene frequency of the normal allele in the population? What percentage of the females in this population would be expected to be normal?

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Chapter 26

Forces Affecting Hardy-Weinberg Equilibrium

Gene and genotypic frequencies remain constant under panmixia unless upset by certain forces. These forces may be grouped as systematic and dispersive forces.

Forces affecting Hardy-Weinberg equilibrium

Systematic Pressures	Dispersive Pressures
Migration Mutation Selection	Genetic drift Inbreeding
Act both in large and small populations	Act only in small populations
Can be predicted in amount and direction	Can be predicted in amount but not in direction

SYSTEMATIC PRESSURES

Here we shall consider a large population so that the effects of systematic pressures are understood in the absence of dispersive pressures.

MIGRATION

Migration means movement. It may be immigration or emigration. Migration may be a factor for a change in the gene frequency in a population. For instance, a farmer has a herd of cattle in which all are horned. Horned condition being recessive to polled condition, that all animals are of pp genotype. Thus the gene frequency of P gene in the population is zero. Let us suppose that he introduces a polled bull (P-) in the herd and as a consequence of mating this bull with the cows in the herd some polled calves are born. Thus this immigrant has increased the gene frequency of P-gene to a frequency greater than zero.

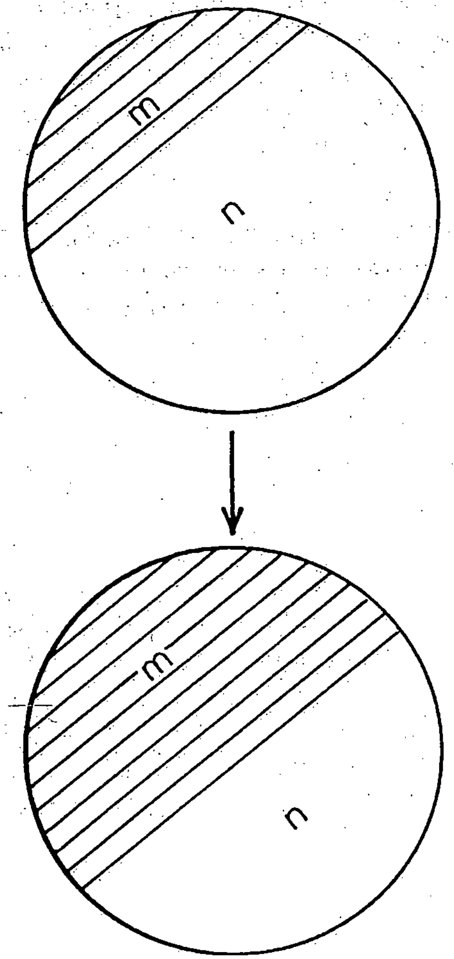
Individuals inhabiting different localities may differ in their gene frequencies because nature selects for different optimal values in different environments. Let us suppose that a large population in a locality consists of a fraction of m of immigrants in each generation from another habitat. The migration results in a mixed population consisting of m population of immigrants and (1-m) proportion of natives. Let q_m be the gene frequency amongst the immigrants, q_o amongst the natives. The gene frequency, q_1 in the mixed population will be

$$\begin{aligned} q_1 &= mq_m + (1\text{-}m)q_o \quad \} \\ &= mq_m + q_o - mq_o \quad \} \\ &= m(q_m - q_o) + q_o \quad \} \end{aligned}$$

The rate of change (Δq) in the gene frequency is

$$\begin{aligned} \Delta q &= q_1 - q_o \\ &= (m\,(q_m - q_o) + q_o) - q_o \\ &= m\,(q_m - q_o) \end{aligned}$$

The rate of change of gene frequency in a population subject to immigration depends on the immigration rate and on the difference in the gene frequency between immigrants and natives.


Migration

MUTATION

The effect of mutation on the genetic properties of the population differs according to whether they are non-recurrent or recurrent mutations. The non-recurrent mutations with no selective advantage for the mutant allele can not produce a permanent change in the population. But the recurrent mutations produce permanent changes in the population since the mutations arrive anew in every generation and generates fresh genetic variability in every generation. For a particular loci, the mutation rate remains reasonably constant from generation to generation under constant environmental conditions.

Let us assume that the wild type gene is 'A' and its mutant is 'a'. Further, let us assume that A mutates to a at the rate of u per generation and a-mutates to A-gene at the rate of v per generation. Suppose if the initial gene frequencies are p(A):q(a), then a-gene may increase up in the next generation by forward mutation and decrease by vq by reverse mutation in the next generation.

Thus the change in q will be

$$\Delta q = up - vq$$

$$\text{(gain) - (loss)}$$

This sort of situation will ultimately lead to an equilibrium gene frequency at which no further change takes place, where the gain in gene frequency equals the loss in gene frequency. The reason is that if the frequency of one allele increases fewer of the other are left to mutate in that direction and more are available to mutate in the other direction. The point of equilibrium can be found by equating the change of frequency, q, to zero, i.e. gain = loss.

Thus at equilibrium

$$u_p = v_q$$

$$u(1-q) = vq \qquad \text{Similarly}$$

$$u - uq = vq$$

$$u = uq + vq \qquad p = \frac{v}{u+v}$$

$$u = q(u+v)$$

$$\frac{u}{u+v} = q$$

The following are the conclusions that can be drawn from the effect of mutation on gene frequency:

1. Mutation is the sudden change in the structure of gene.
2. Mutation from wild type to mutant is the forward mutation and from mutant to wild type is the reverse mutation or backward mutation.
3. The equilibrium value is entirely determined by the opposing rates of mutation (u & v) and not by the initial gene frequencies.
4. The mutation rates u & v are constant for a particular locus and therefore the equilibrium attained is stable.
5. However, the mutation rates are generally low ranging from 10^{-4} to 10^{-8} per generation. With such low rate of mutation their effects in changing the gene frequency is so feeble that other forces such as selection, migration and sampling variation from generation to generation are almost certain to overwhelm the mutation effects in determining the gene frequencies. The effect of mutation is very feeble. Thus if u and v are the only forces acting, it is of little practical consequence.
6. Reverse mutations are generally less frequent than forward mutations. Studies indicate that reverse mutations are one tenth as frequent as forward mutations. Thus the equilibrium frequencies arising from mutation alone would yield 0.1 wild type gene and 0.9 mutant gene as shown below:

$$p = \frac{v}{u+v} = \frac{0.1}{0.9+0.1} = 0.1;\ q = 0.9$$

That is the mutant genes will be common and wild rare. But this is not a situation we come across in natural population and in fact a reverse condition exists. Hence, it is clear that selection may operate against the mutant types and it is responsible for rarity of mutant genes seen in natural population. It is also well known that many mutations are generally deleterious.

7. Mutation rates can be accelerated by ionizing radiation, high temperatures, X-ray and other short rays, chemicals, etc. Any locus at which the gene frequencies are at equilibrium, from the opposing effects of mutations alone, will not be affected by a change of mutation rate, provided the change in the environment affects both forward and reverse mutations proportionately.

SELECTION

Selection is choosing the parents of next generation. So far we have assumed that all the individuals irrespective of their genotypic make up constitute equally to the next generation. It is a common place to see individuals differ in their fertility, mating ability and viability and consequently they may contribute differing number of offsprings to the next generation. The proportionate contribution of offspring to the next generation is called the fitness (W) of the individuals or sometimes referred to as adaptive or selective value. The differences in fitness may be associated with the genotypes of the individuals. Thus differences in the contribution of offsprings to the next generation by different genotypes lead to a change in the gene frequency and consequently genotype frequencies. The strength of selection can be expressed, as selection coefficient symbolised by s, which is the proportionate reduction in the gametic contribution of a particular genotype compared with a standard genotype, usually the most favoured one. The contribution of a favoured genotype is then taken as one (w=1) and that of the genotype selected against is taken as 1-s (w=1-s). For instance, if the selection coefficient, s = 0.1; this means that for every 100 zygotes produced by the favoured genotypes only 90 are produced by the genotype selected against. In nature recessives are generally deleterious, in other words their phenotypes are not optimal for their well being. It is reasonable to assume that the fitness of the genotypes is dependent on the degree of dominance shown by the genes with respect to fitness.

Let us assume that there is partial selection and that the selection coefficient against a genotype is s. The fitness (w) of the aa genotype is then (1-s). When there is no dominance the selection coefficient against heterozygotes (Aa) will be exactly intermediate, i.e. 1/2s. Therefore the fitness, (w) of heterozygotes will be (1-1/2s). The homozygotes AA will be the most favoured one and hence will have a fitness value w = 1. The same may be diagrammatically represented as

Genotype	:	AA	Aa	aa
Fitness (w)	:	1	$(1-\frac{1}{2}s)$	(1-s)

When there is complete dominance, the fitness of genotypes will be:

Genotype		AA	aa
		Aa	
Fitness (w)	:		
			(1-s)

Note that dominants whether homozygous or heterozygotes show no difference in fitness.

In overdominance, the heterozygotes are most favoured compared to both the homozygotes and hence their fitness:

		aa	AA	Aa
Genotype fitness (w)	:	$(1-s_2)$	$(1-s_1)$	1

Partial selection against recessives:

Here the dominants are favoured and may be due to higher fertility and viability compared to recessives.

Assume that there is complete dominance. Let us further assume that for every offspring produced by dominants, (1-s) offsprings are produced by recessives, where s is the selection coefficient against recessives. Since the population is in Hardy-Weinberg equilibrium in each generation before selection, we shall assume an initial genotype frequency of (p^2, 2pq, q^2) for the population. The change in the gene frequency after one generation of selection may be worked out as:

Genotype	AA	Aa	aa	Total
Initial frequency	p^2	2pq	q^2	1
Fitness (w)	1	1	1-s	
After selection	p^2	2pq	$q^2(1-s)$	$1-sq^2$

Frequency in the selected population $\frac{p^2}{1-sq^2}$ $\frac{2pq}{1-sq^2}$ $\frac{q^2(1-s)}{1-sq^2}$

The a-gene frequency in the selected population is

$$\frac{pq}{1-sq^2} + \frac{q^2(1-s)}{1-sq^2} = \frac{pq + q^2(1-s)}{1-sq^2}$$

$$\frac{q(p+q-sq)}{1-sq^2} = \frac{q(1-sq)}{1-sq^2}$$

The gene frequency in the subsequent generation obtained from random mating between selected individuals is the same as that in selected population, i.e.

$$q_1 = \frac{q(1-sq)}{1-sq^2}$$

Therefore when there is complete dominance and partial selection against recessives, the change in gene frequency is

$$\Delta q = q_1 - q$$

$$= \frac{q(1-sq)}{1-sq^2} - q$$

$$= \frac{q(1-sq) - q(1-sq^2)}{1-sq^2} \qquad \frac{q - sq^2 - q + sq^3}{1-sq^2}$$

$$= \frac{-sq^2(1-q)}{1-sq^2}$$

When there is no dominance the fitness values for the three genotypes will be AA 1 : Aa 1-1/2s : aa 1-s as pointed out earlier. The changes in the gene frequency in one generation can be demonstrated as

$$\Delta q = \frac{-1/2sq(1-q)}{1-sq^2}$$

Note that the minus sign for q in the above equations indicate that the a-gene frequency decreases when aa genotype is selected against whether there is dominance or not. This reduction in gene frequency proceeds until it reaches a low level where the recessive gene is only supported by mutation. It is reasonable to attribute the low frequency of deleterious genes (recessive genes) that we find in natural populations to the balance between mutation tending to increase the frequency and selection favouring to decrease it.

Complete elimination of recessives: Recessives get eliminated completely when the recessives are lethal or unwanted in artificial selection. This means that selection coefficient against recessive genotypes is one. Hence the fitness of genotypes are AA 1: Aa 1 : aa 0.

Starting with an initial population of (p^2, $2pq$, q^2) as before we could demonstrate that $q_1 = \frac{q}{1+q}$ as shown below:

Genotypes	AA	Aa	aa	Total
Frequency	p^2	$2pq$	q^2	1
Fitness	1	1	0	
After selection	p^2	$2pq$	0	$p(1+q)$
Frequency in selected population	$\frac{p^2}{p(p+q)}$	$\frac{pq}{p(1-q)}$		

$$p^2 + 2pq$$
$$p(p + 2q)$$
$$p(p + q(q)$$
$$p(1 + q)$$

$$q_1 = \frac{pq}{p(1+q)} = \frac{q}{1+q}$$

Where q is the initial gene frequency.

Let it be assumed as q_o, then we get

$$q_1 = \frac{q_o}{1 + q_o}$$

The gene frequency after the second generation when recessives are completely eliminated.

$$q_2 = \frac{q_1}{1 + q_1} = \frac{q_1}{1 + 2q_0}$$

In general the gene frequency after n generations of complete elimination of recessives is

$$q_n = \frac{q_o}{1 + nq_o}$$

Conversely the number of generations required to change the gene frequency from q_o to q_n under complete elimination of recessives is

$$n = \frac{1}{q_n} - \frac{1}{q_o}$$

The equation can be derived as below:

$$q_n = \frac{q_o}{1 + nq_o}$$

$$q_n + nq_nq_o = q_o$$

$$n = \frac{q_o - q_n}{q_n \quad q_o} = \frac{1}{q_n} - \frac{1}{q_o}$$

Where n = number of generations

q_n = the desired frequency of gene at nth generations.

q_o = the initial frequency of gene before selection.

EXERCISE

1. What are the forces affecting the Hardy-Weinberg equilibrium?
2. What are differences between the systematic and dispersive forces?
3. Discuss on the influence of migration on Hardy-Weinberg equilibrium.
4. Discuss on mutation and Hardy-Weinberg equilibrium.
5. Define the following
   a) Selection
   b) Selective or Adaptive value
   c) Selection co-efficient
6. Give briefly on the influence of dominance on the fitness of the genotype.
7. Describe on the change in gene frequency due to partial selection against recessives.
8. Derive the formula for estimating the number of generations required to change the gene frequency under complete elimination of recessives.
9. In a population suppose the mutation rate from A to a is 0.005 per generation and that to A is 0.0005 per generation, find the equilibrium value of q.
10. If u = 0.005 and v = 0.0015, what will be the equilirbium values of gene frequencies of p and q?
11. Among pure herd of Holstein, about one calf in 256 calves born with red spots rather than black. Red is recessive and if none of them reproduce what would be the proportion of calves with red spots in the next generation and after 10 generations?
12. Two alleles exist in a population in equal frequency p = q = 1/2. Show how many generations would be required to reduce the frequency of

q from 1/2 to 1/5 if the homogygous recessive suddenly become lethal.

13. The present frequency of albinism is 1 out of 20,000. How long would it take to reduce the gene frequency to half of its value, when albinism is totally eliminated?

14. Suppose a recessive trait occurs 1/1000 of a random mating population. How many generations of complete selection against recessive individual would it need to reduce the proportion to 1/10,00,000?

15. One in 2500 individuals have an abnormality due to homozygous recessives, What proportion will have abnormality after 50 generations of complete elimination?

Chapter 22

Dispersive Forces

So far we have been discussing the maintenance of Hardy-Weinberg equilibrium in large populations. In reality, populations are finite and not large enough to discount chance of fluctuations in gene frequency. In small populations the property of constancy of gene and genotypic frequencies no longer hold good since they are subject to random fluctuation arising from sampling of gametes. Gametes carry a sample of genes from the parent population to the offspring population. If the population is small, the gene frequency is bound to differ from generations to generation due to errors of sampling. Smaller the population, the greater are the errors of sampling. The random change in the gene frequency from generation to generation arising out of sampling error is known as dispersive process. Hence dispersive process are of consequence only in small populations, while they are feeble enough to be ignored in large population.

The most important consequences of dispersive process are genetic drift and inbreeding.

GENETIC DRIFT:

Let us assume that a population is being maintained with no overlapping generations and with a constant strength of N diploid individuals in each generation. The N diploid individuals in any generation is a product of union of 2N gametes drawn at random from the previous generations. Let q_o be the gene frequency in the initial population. The population being small the gene frequency in the next generation, q_1 may fluctuate by pure chance according to the bionomial expansion.

$$(P_o + q_o)^{2N}$$

That is, there may be (2N + 1) possible values for q_1.

$$\text{viz.} \quad o, \quad \frac{1}{2N}. \quad \frac{2}{2N}, \quad \cdots\cdots\cdots \quad \frac{2N-1}{2N}, \quad 1$$

In other words q_2 will increase or decrease at random as a result of binomial sampling, the direction of which is unpredictable, but the magnitude of change or shift (δq) from the parental generation gene frequency can be stated in terms of variance of the change in q.

$$\delta^2 \; (\delta q) \quad = \frac{p_o q_o}{2N}$$

In the second generation, the sampling process is repeated. But this time it starts from a different gene frequency i.e. q_1 and so the second sampling leads to further dispersion. The effect of sampling through successive generations is that the gene frequency fluctuates randomly and drifts away from the initial values. This drifting of gene frequency resulting from sampling process in small populations is known as genetic drift.

As the drift proceeds the variance of the gene frequency increases. Letting q_t the frequency of a-allele in generation t,

the expectation of $q_t = E (q_t) = q_o$

and the variance of $q_t = 2 (q_t) = p_o q_o \; (1\text{-}1\text{-}1/2N^t)$

The above results are most easily interpreted if we imagine a very large population subdivided into a large number of isolated random mating groups (lines) of N diploid individuals at random. All the lines initially have the same base population gene frequency, q_o. But as generation passes, the gene frequency of the lines fluctuates due to random sampling in successive generations and become differentiated.

Then the equation $E (q_t) = q_o$

states that the mean gene frequency (averaged over all lines) remains unaltered.

Random fluctuation of the gene frequency is only possible so long as its frequency in the previous generation is between zero and one, but not 0 or 1. As a result of random drift the gene frequency may ultimately reach either 0 or 1 and until then drift continues. When a particular gene reaches a frequency of 1, it is said to be fixed and when it reaches o, it is said to be lost. When gene frequency is 1, all the individuals are homozygotes in that line. The proportion of the lines in which different alleles of a locus are fixed is equal to the initial frequencies of the allele, i.e., if the initial frequencies of the alleles, A and a, are p_o and q_o respectively, the A-allele will be fixed in po proportion of the total lines and the a-allele inthe remaining qo of the lines. The variance of the gene frequencies among the lines will then be $p_o q_o$ which can be verified by finding the limiting value of 2 (q) by letting the t as being equal to infinity.

INBREEDING

Dispersive process leads to increase in homozygosity with a corresponding decrease in heterozygosity. The reason is that the dispersible process forces the gene frequencies from intermediate values towards the extreme values. Heterozygotes are most numerous at intermediate gene frequencies. As an illustration we shall estimate the heterozygotes frequencies when

q =	0.9	0.5	0.1
H = 2pq	0.18	0.5	0.18

We find that the frequency of heterozygotes is maximal when p = q = 0.5. Hence the drift of gene frequencies to extreme (low or high) values leads, on an average, to a decline in the frequency of heterozygotes with a corresponding increase in both homozygotes.

We shall look at the dispersive process in terms of inbreeding. Inbreeding means the mating together of individuals that are related to each other by ancestry. It is obvious that in a bisexual population, an individual has two parents, 4 grand parents, 8 great grand parents and 2^n ancestors in n generations back. Therefore, any pair of individuals in a population may be related to each other in a more or less remote past. The smaller the size of the population, the greater is the number of common ancestors and less remote are the common ancestors. Thus individuals mating at random in small populations are closely related and the resulting properties are the consequences of inbreeding, i.e., increase in homozygosity at the expense of heterozygotes.

Estimation of inbreeding populations: The inbreeding can be measured by means of inbreeding coefficient (F). The inbreeding coefficient may be defined as the probability that the two alleles of any locus in a diploid organism are identical by descent.

The concept of identity by descent was developed by Male'cot (1948). Two genes of a locus may be identical for two exclusive reasons.

Identical in state **:** Two alleles may be both A as may be seen in homozygotes. Since they are alike in function they are called alike in state. ***Identical by descent*** **:** Two alleles are said to be identical by descent (or simply identical) if one of them have been derived by direct replication from the other or if they are both copies of the same gene in a common ancestor. For example, a parent is of the genotype, X_1, X_2 (where X_1 X_2 may represent any of the three genotypes, namely, AA, Aa and aa). It passes one of its two genes say X_1 to its offspring and its offspring is of the genotype X_1 X_3. Now X_1 of the offspring and that of the parent are said to be identical by descent since the former was derived by direct replication of the latter. Suppose the same parent passes another replicate of its X_1 to yet another offspring and the second offspring is of the genotype X_1 X_4. The X_1 of the two offsprings (sibs) are said to be identical by descent since the two X_1 genes are copies of the same gene occurring in their common ancestor. Thus identity by descent provides a measure of inbreeding through the degree of relationship between the mating pairs.

Let us assume that a population is being maintained with no overlapping generations, with random mating, including selfing in random amount, absence of systematic processes and with a constant size of N diploid individual in each generation. Assume that the population is a hermophrodite marine organism shedding equal number of gametes into the sea. Assume that all the genes are of independent origin in the base population. Thus based on identity by descent the gametes shed can be viewed as belonging to 2N different sorts. The gametes of any one sort carry identical genes and those of different sorts carry genes that are independent in origin. The probability that a pair of gametes taken at random being identical is 1/2N. In other words, any gamete has (1/2N)th chance of uniting with another of the same sort. Since the inbreeding coefficient is the probability that the two alleles of a locus are identical by descent, it follows that the average inbreeding coefficient of the population in the next generation will be 1/2N.

Therefore the rate of inbreeding is $F = 1/2N$

To deduce the above equations with ease we have considered a simplified population of hermaphrodite with N breeding individuals in each generation. We shall now consider a model which allows for separate sexes and differing number of breeding individuals in each generation so that the above equation may be modified to estimate drift in realistic populations.

The only difference is the use of effective population size, Ne in the place of N in the equation. The effective population size is the number of individuals that would give to sampling variance and rate of inbreeding appropriate to the conditions under consideration, if they were bred in a manner of the simplified population described.

The rate of inbreeding with the effective population size is $F = 1/2Ne$.

Where Ne - is the effective population size.

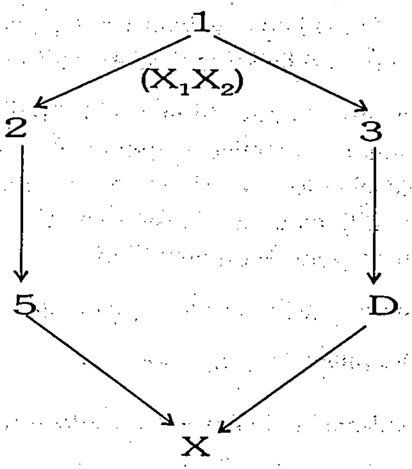
Estimation of inbreeding in pedigreed population:

Inbreeding coefficients of individuals may be estimated when their pedigrees are available. This method overcomes the difficulty of estimating inbreeding coefficients in the population if there are overlapping generations. In the breeding of farm animals, it is a common experience to find the same animal being used as a parent in two or more generations. Another advantage is that inbreeding may be measured by this method even if there are assortative matings (selection).

As already stated, the inbreeding coefficient is the probability that the two alleles of any locus in a diploid organism are identical by descent. In has two ...

...coefficient of an individual is the probability of ...ed by the gametes, that united and produced the ...ntical by descent. Consider the pedigree represented in ...s the individual for which the inbreeding coefficient is to be ... and S and D are his sire and dam and 1 is their common ancestor. ...want to estimate the probability of X receiving genes identical by descent through their parents from the common ancestor, namely, 1. Let the genotype of 1 be (X_1 X_2) and consider its two offsprings namely 2 and 3. The probability that the individuals 2 and 3 both receive X_1 gene from their parent 1, is 1/4 and the probability that both receive X_2 genes is again 1/4. Therefore the total probability that 2 and 3 receive identical genes (identical by descent) is 1/2 and the probability that they receive different genes is 1/2. If they receive different genes from their parent 1, the probability of these different genes being identical by descent as a result of previous inbreeding undergone by the common ancestor 1, is $1/2F_A$. Where F_A is the inbreeding coefficient of the common ancestor, namely the parent, 1. Therefore the total probability that individuals 2 and 3 receive genes identical by descent is $1/2 + 1/2F_A = 1/2\ (1 + F_A)$. In other words, this is the probability that two gametes taken at random from the parent 1 contain genes identical by descent. Now consider the rest of the paths to X through sire and dam. The probability that the individual 2 passes on the genes it got from 1 to S is 1/2 and from S to X is 1/2. Similarly, looking at the dam side, the probability that the genes of 1 received by 3 is passed on to D and from D to X is 1/2 in each case. Putting all these together, we find the probability that X receives identical genes from the common ancestor is

Inbreeding Coefficient



1 - Common Ancestor

X - Individual - who have inherited genes identical from common ancestor through sire and dam

कृपया रेषा मारुन
पुस्तक खराब करु नये.

$$Fx = 1/2\ (1\text{-}F_A)\ (1/2)\ (1/2)\ (1/2)\ (1/2)$$

In general we may write

$$F_x = 1/2\ (1\text{-}F_A)\ (1/2)^n$$

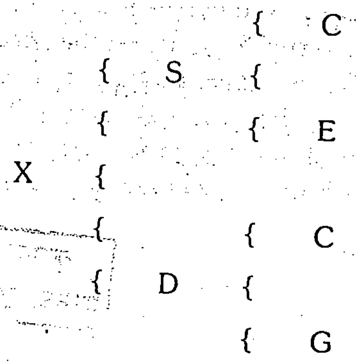
where n is the sum of the number of generations back to common ancestor from one parent and the number of generation from the common ancestor to the other parent. As there may be several common ancestors, the contribution from each common ancestor is summed over to give the inbreeding coefficient of the individual. Therefore, on realignment of the equation, we get

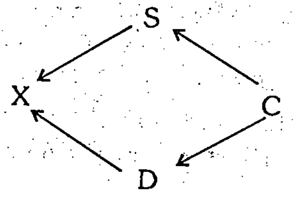
$$Fx = 1/2 \quad \sum \{(1/2)^n\ (1 + F_A)\}$$

Estimation of inbreeding coefficient in individuals:

As a first step draw an arrow diagram of the pedigee to facilitate estimation of inbreeding coefficient. Let X be the individual for which the inbreeding coefficient F is to be determined and S and D are its parents. Draw arrows from S to X and from D to X with the points of arrows directed towards X. Locate the first common ancestor. A common ancestor is one who is related to both sire and dam of the individual. Draw arrows from the common ancestor to sire and dam with the arrows passing through intervening ancestors if any and always pointing towards S and D. Remember that there may be more than one pathway through which a common ancestor may be related to S and D. Locate all the rest of the common ancestors and connect them to S and D by arrows as stated above. Also remember, that the same common ancestor do not appear in more than one place in the arrow diagram.

Example:





The probability that the two individuals S and D receive the same gene from the common ancestor is 1/2 and the probability that the same gene is transmitted to X is 1/2. Then the inbreeding coefficient is estimated by the formula.

$$Fx = \sum (1/2)^{n_1+n_2+1}$$

Where 1/2 = halving process

n_1 = no. of generations from the common ancestor to the father

n_2 = no. of generations from the common ancestor to the mother

The Fx in the above example can be calculated as below:

Common ancestor	Path	n_1	n_2	Contribution
C	S-C-D	1	1	1 $(1/2)^{1+1+1}$
				$(1/2)^3 = 1/8 = 0.125$

When the common ancestor itself is inbred the inbreeding coefficient can be estimated using the following formula.

$$Fx = \sum (1/2)^{n_1+n_2+1} (1+FA)$$

Where FA is the inbreeding coefficient of the common ancestor.

EXERCISE

1. Describe on the consequences of genetic drift on the properties of small population.
2. Give an account on inbreeding and its effect on the gene frequencies of the population.
3. Define inbreeding coefficient and discuss on its estimation.
4. Define effective population size and what is the rate of inbreeding with the effective population number.
5. Differentiate between identical by state and identical by descent.
6. Give the formula for the estimation of inbreeding coefficient of the individual and its derivation.
7. Find out the expected rate of inbreeding in a population of Drosophila maintained by 20 pairs of parents in each generation.
8. Given 300 breeding females and 50 breeding males in a random mating group. What is the effective size and rate of inbreeding?
9. Given 280 breeding females and 40 males in a random mating group. What is the effective population number and estimate rate of inbreeding?
10. If N_f=1000, N_m=40 find out Ne.
11. Given 4 generations of a population, the effective sizes are 30, 90, 700 and 6000.
12. In four generations, the population sizes are 20, 100, 500 and 5000. What is the average effective population size?
13. Given 4 generations with effective numbers 20, 100, 800 and 5000, What is the average effective sizes?
14. Compute Fx in the following pedigree:

```
          {  C
    {  S  {  E
X   {     {  C
    {  D  {  G
```

15. Compute Fx in the following pedigree:

```
          {  C
    {  S  {  E
    {
X   {     {  C
    {  D  {  E
```

16. Compute Fx in the following pedigree:

```
          {  C
    {  S  {  H
X   {     {  B
    {  D  {        {  C
             E     {
                   {  H
```

17. Compute Fx in the following pedigree:

```
                   {  4
          {  1     {
          {        {  5
          {
    {  S  {  2     {  4
    {              {  5
X   {     {  S
       D  {
          {  3
```

18. Compute Fx in the following pedigree:

```
                   {  F
          {  C     {
          {        {  G
          {
    {  A  {        {  F
    {     {  D     {
    {              {  H
X   {     {  C
    {  B  {
          {  E
```

Chapter 28

Values

The genetic properties of a population are expressed in terms of gene frequency and genotype frequency. To understand the connection between these properties on one hand and the quantitative differences on the other hand, a new concept is introduced which is known as value.

The observed value of an individual is called phenotypic value of that individual. All observations like mean, variance, covariance etc. are based on the phenotypic values. The observed phenotypic value is affected both by the genotype and environment. Hence the phenotypic value may be partitioned into components attributable to the influence of genotype and environment. The components of the value associated with genotype is called genotypic value and that associated with the environment as Environmental deviation.

It may be written symbolically as

$$P = G + E$$

Where P = Phenotypic value

G = Genotypic value and

E = Environmental deviation

Environmental Deviation:

Environmental deviation may represent differences between individuals of the same genotype raised under similar conditions. Environmental variations reduce precision in genetic studies. The experimenter can minimize the environmental variations by careful management or by proper design of experiments.

The environmental deviations are due to the following two sources:

1) Differences in external environments

2) Variability in the internal environment of the organism.

Nutrition and climate are the common external causes of environmental variation. Maternal effects form another source of external environmental variation. Mothers may differ in their pre and post-natal influences on their offsprings and which may be mainly nutritional. Another reason for the external variation is errors in measurements which are usually negligible, unless the trait is measured by subjective evaluation such as visual grading of carcass.

Internal environment variability is due to errors in development. The two sides of Drosophila may differ in their bristle number which might be due to local developmental accidents rather than due to differences in external environment to which both sides of the fly were subjected to in the larval or adult stage.

Genotypic value:

Genotypic value may be measured by taking a mean of a large population with the same genotype raised under similar conditions. It will be assumed that any environmental deviation uncontrolled by man has been randomized and the mean environmental deviation as a whole for the population is taken as zero, so that the mean phenotypic value is equal to the genotypic value.

The genotypic value may be partitioned into components representing additive gene action, dominance and epistasis. It may be written symbolically as

$$G = A + D + I$$

Where G is the genotypic value

A is the additive value

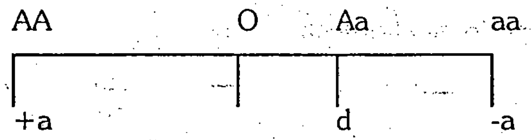
D is the dominance value and

I is the interaction or epistatic value

When the mean environmental deviation is taken as zero then the mean phenotypic value is equal to the mean genotypic value.

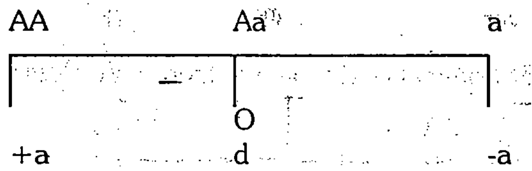
Scale of genotypic value:

Genotypic value can be measured by means of scale of genotypic value. Arbitrary values are assigned to the genotypes considering a single locus with the two alleles A and a, the genotypic value of one homozygote AA is assigned as +a and of the other homozygote aa as-a and that of the heterozygote Aa as d. The origin or point of zero value is between the midway of the values of the two homozygotes.

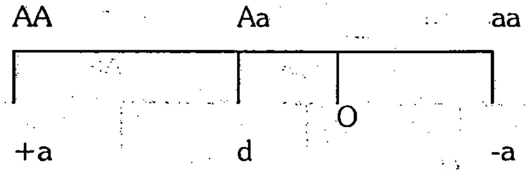


The d value depends on the degree of dominance.

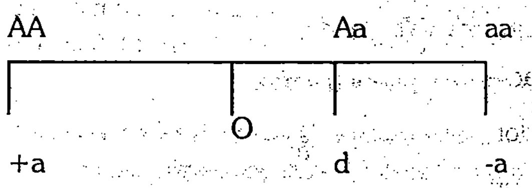
a) If there is no dominance d=o



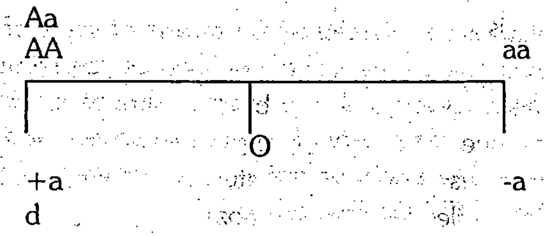
b) If A is dominant, d is positive



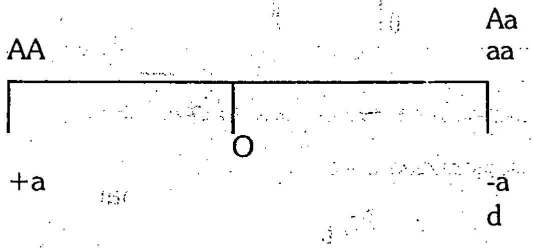
c) If a is dominance over A, d is negative



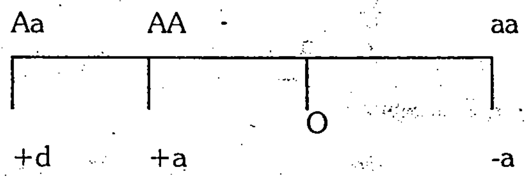
d) If A is complete dominance then d = +a

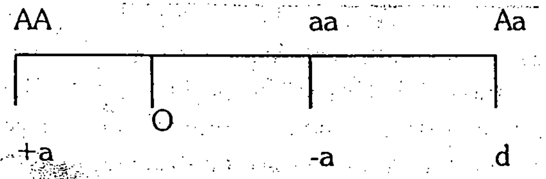


e) If a is complete dominance then d = -a



f) If there is over dominance d is greater than +a or lesser than -a.





The degree of dominance is expressed as d/a.

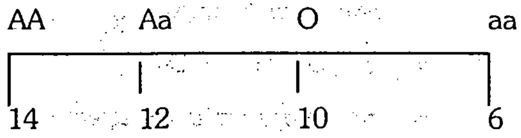
Example: There is a dwarfing gene pg in mouse known as pygmy. This gene reduces the body size. It was present in a strain of mice maintained by MacArthur. The weights of three genotypes at 6 weeks of age are approximately as follows:

Normal : 14 G

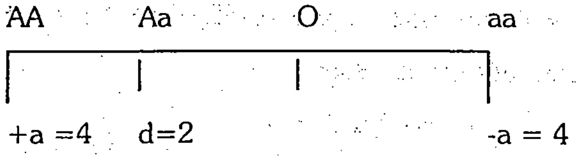
Heterozygous normal : 12 G

Recessive (pygmies) : 6 G

The scale of genotypic value is drawn as shown below. The average weights are the genotypic values.



The mid point in genotypic value between the two homozygotes is 10g and this is the origin.



The degree of dominance = d/a = 2/4 = 1/2 = 0.5 = 50%

Population mean:

The population mean refers equally to the phenotypic or genotypic values. It is the mean gene frequency of the character in the population. Let the gene frequencies of the two alleles A and a be p and q respectively. The values and the genotypic values will be as given below. The mean value in the whole population is obtained by multiplying the value of each genotype by its frequency and adding up the three genotypes.

Genotype	Frequency	Value	Mean value Freq x value
AA	p^2	+a	p_2a
Aa	2pq	d	2pqd
aa	q^2	-a	$-q_2a$

$$\text{Total} = a\,(p-q) + 2dpq$$

Mean values are summed over and simplified.

$$\begin{aligned}\text{Population mean} &= p^2a + 2pqd - q^2a \\ &= p^2a - q^2a + 2pqd \\ &= a\,(p^2 - q^2) + 2pqd \\ &= a\,(p+q)\,(p-q) + 2pqd\end{aligned}$$

This is both the mean genotypic value and the mean phenotypic value of the population with the respect to the character. The contribution of any locus to the population mean has two terms.

a (p-q) attributes to homozygotes and 2pqd attributes to heterozygotes.

If there is no dominance, then

$$d = o \text{ and } 2pqd = o$$

$$\text{Population mean} = a\,(p - q)$$

It there is complete dominance then $d = a$

Population mean = square of the gene frequency

$$\begin{aligned}\text{Population mean} &= a\,(p-q) + 2pqa \\ &= ap - aq + 2pqa \\ &= a\,(p - q + 2pq) \\ &= a\,(p+2pq-q) = a\,(p-q)\,(1 + 2q)\} \\ &= a\,(p(1+2q) - q) = a\,\{(1 - q - q)\,(1 + 2q)\} \\ &= a\,(1 - 2q_2)\end{aligned}$$

In the example of pygmy mice, the population mean

$$= a(p-q) + 2pqd$$

$$p = 0.9\ q = 0.1\ a=4\ d=2$$

$$= 4x(0.9-0.1) + 2x0.9x0.1x2$$

It should be measured from mid point

$$\text{Population mean} = 10+3.56 = 13.56$$

Average effect:

The transmission of value from parent to offspring can be dealt with not by means of genotypic values alone, because parents transmit their genes and not their genotypes to the next generation. Genotypes are created fresh in each generation. A new measure of value is therefore needed which will refer to genes and not the genotypes. The new measure is the average effect. It is the mean deviation from the population mean of the individuals. For example if the number of gametes carrying A unite at random with the gametes from the population, then the mean deviation from the population mean of the genotypes so produced is equal to the average effect of the gene A. The averge effect of a gene depends on the gene frequency, and the average effect is a property of the population as well as of the gene.

The genotypic values of homozygotes and heterozygotes, a and d by which population mean is expressed are related to the average effect. Consider a locus with the two alleles A and a, at the frequencies of p and q respectively. The average effect of A gene is symbolised as X. If gametes carrying A unite at random with gametes from the population, the frequencies of the genotypes produced will be p of AA and q of Aa. The genotypic value of AA is +a and that of Aa is d, and the mean of these, is pq + qd. The difference between this mean value and the population mean is the average effect of the gene A.

$$\begin{aligned}\alpha\,1 &= pa + qd - \{a(p-q) + 2pqd\} \\ &= q\,(\{a+d\,(q-p)\}\end{aligned}$$

The average effect of the gene a is similarly = X2

$$\alpha\,2 = -p\{a+d\,(q-p)\}$$

Breeding value:

Additive gene effects are connected with the breeding values of an individual since the parents pass on their genes and not their genotypes to their offspring. The average effects of parents' genes determine the mean genotypic value of its progeny. The value of an individual determined by the mean value of its progeny is called the breeding value of the individual. It is equal to the sum of the average effects of the gene it carries, the summation being made over the pairs of alleles at each locus and over all loci controlling the trait. Thus additive gene effects are also referred to as breeding value. In a population under Hardy-Weinberg equilibrium mean breeding value must be zero or mean breeding value must be equal to the mean genotypic value and to the mean phenotypic value.

Dominance deviation:

If a single locus alone is under consideration, the difference between the genotypic value G and the breeding value A of a particular genotype is known as the dominance deviation. The genotypic value can be partitioned as

$$A = A+D$$

Deviation of the genotypic value from the breeding value give rise to dominance deviation. Thus dominance deviation may arise if the alleles of the same locus interact and show the property of dominance. When this concept is extended to a multilocular case, the dominance deviation is the sum of the dominance deviation of the individual locus.

Interaction deviation:

Metric traits are polygenic in inheritance and hence determined by many loci. Their genotypic value will then contain in addition to breeding value and dominance deviations, the interaction or epistatic effects caused by intraocular interactions. Thus the phenotypic value of a metric trait can be partitioned into

$$P = G+E$$

Where $G = A+G+I$

EXERCISE

1. Define phenotypic value and what are its components.
2. Describe on the components of environmental deviation.
3. Discuss on the genotypic value.
4. Write short notes on scale of genotypic value.
5. Write short answers on the following:
   a) Population mean
   b) Average effect
   c) Breeding value
   d) Dominance deviation
6. Draw the scale of genotypic value when the population sizes are

AA	Aa	aa
28	24	16

and find out the degree of dominance

7. Draw the scale of genotypic value when the population sizes are as below

AA	Aa	aa
14	12	6

Estimate the degree of dominance and the population mean when q is 0.2

8. Mean sizes of 3 genotypes in a population are as follows:

AA	Aa	aa
32	24	12

1) Draw the scale of genotypic values.
2) What is the degree of dominance?
3) What is the population mean when q=0.2?

9. Determine the population mean using the following informations:

Genotypes	MM	Mm	mm
Number of animals	375	300	75
Genotypic values	200	238	606

Chapter 29

Variance

The individuals differ in their metric characters. The amount of variation is measured and expressed as the variance. Corresponding to the partition of phenotypic value into genotypic value and environmental deviation, P=G+E, the phenotypic variance (V_P) is partitioned into genotypic (V_G) and environmental variances (V_E).

$$V_P = V_G + V_E$$

The genotypic variance is the variance of genotypic value and the environmental variance is the variance of environmental deviations. The total variance is the phenotypic variance.

The components of variance and the values whose variance they measure are listed below:

Variance component	Symbol	Value whose variance is measured
Phenotypic	V_P	Phenotypic value
Genotypic	V_G	Genotypic value
Additive	V_A	Breeding value
Dominance	V_D	Dominance deviation
Interaction	V_I	Interaction deviation
Environmental	V_E	Environmental deviation

The total variance is then the sum of these components.

$$V_P = V_G + V_E$$
$$V_G = V_A + V_D + V_I$$
$$\text{Hence } V_P = V_A + V_D + V_I + V_E$$

Measurement of variance:

The observed phenotypic variance is the sum of genetic and environmental variances. Since the genotype and the environment are the only two determinants of the phenotype, if one of the two components is eliminated, the other is measurable. In practice it is not possible to eliminate environmental variance. Even though the environment is uniform, there may be still certain amount of micro-environmental variation, which can not be recognised and controlled. However genetic variation can be eliminated by inbreeding. A strain inbred for more than 20 generations by brother sister mating is, for all practical purposes, free of genetic variability. Then within the strain, variation is purely environmental. However inbreds are more susceptible to environmental differences. The observed variance of the individuals of a genetically variable strain is the sum of the genetic variance in that strain and environmental variance substraction of environmental variance, estimated by cross-bred individuals therefore gives an estimate of genetic variance.

Example:

In Drosophila, the length of the thorax (in units of 1/100 mm) was recorded as a measure of body size. The phenotypic variance was measured in a genetically mixed population and in a genetically uniform population.

The first estimates the genotypic and environmental variance together and the second estimates the environmental variance alone, so by substraction genetic variance is obtained.

Population	Components	Observed variance
Mixed	$V_G + V_E$	0.366
Uniform		V_E 0.186
Difference	V_G	0.180

The degree of genetic determination

$$= \frac{V_G}{V_P} = \frac{0.180}{0.366} = 49\%$$

This shows that 49% of the total variation of thorax length in the genetically mixed population is attributable to the genetic differences between individuals and 51% to non-genetic differences.

Genotypic variance:

The genotypic variance is divided according to the division of genotypic value into breeding value, dominance deviation and interaction deviation.

Genotypic value $G = A + D + I$

Genotypic variance $V_G = V_A + V_D + V_I$

Genetic variation is due to two aspects. One part is made up of additive effects of genes. This component of variance is called as additive genetic variance. Another part of variation is made up of non-additive effect of genes. These are non-additive variance, and the causes are dominance and interaction. Additive gene effects are connected with the breeding values of the individual, since the parents pass on their genes and not their genotypes to their offspring.

The variances are obtained by squaring the values, multiplying by the frequency of the genotype concerned and summing over the three genotypes, AA, Aa and aa.

Genotypes	AA	Aa	aa
Breeding value	2qx	(q-p)x	-2px
Genotypic frequency	p^2	2pq	q^2

$$\begin{aligned} V_A &= (2qx)^2p^2 + (q-p)^2x^2 2pq + (-2px)^2q^2 \\ &= 4p^2q^2x^2 + 2pq(q-p)^2x^2 + 4p^2q^2x^2 \\ &= 4p^2q^2x^2 + 2pqx^2(q^2-2pq+p^2) + 4p^2q^2x^2 \\ &= 4p^2q^2x^2 + 2pq^3x^2 - 4p^2q^2x^2 + 2p^3qx^2 + 4p^2q^2x^2 \\ &= 2pqx^2(2pq+q^2 - 2\cancel{p}q + p^2 + 2\cancel{p}q) \end{aligned}$$

$= 2pqx^2 (p^2 + 2pq + q^2)$

$=$ Since $p^2 + 2pq + q^2 = 1$

$= 2pqx^2$

$= 2pq\ (a+d\ (q-p))^2$

Similarly the dominance variance is

$V_D \ = \{2pqd\}^2$

thus the total genotypic variance

$V_G \ = 2pq\ \{a+d\ (q-p)\}^2 + \{2pqd\}^2$

Properties of genotypic variances:

1) The genotypic variance remains maximum at the intermediate gene frequency. Genes contribute much more variance when at intermediate frequencies than when at high or low frequencies.

2) When a random mating population is in equilibrium, the additive variance arising from all loci together is the sum of the additive variances attributable to each locus separately. The dominance variance is similarly the sum of the separate contributions. If interaction deviations are present, they give rise to another component of variance, the interaction variance.

Environmental variance:

This includes all variations of non-genetic origin. Environmental variance is the source of error that reduces the precision in genetic studies. The causes of environmental variance are nutrition, climate, errors in measurements etc. Maternal effects form another source of variation, which is important particularly in mammals.

Genotype and environment interaction:

We have defined the genotypic value as the mean value of a large group of individuals with identical genotypes raised under exactly similar conditions. In this it was assumed that any environmental variation unintended or uncontrolled by the experimenter is randomized. This environmental variation is known as micro-environmental variation. But the experimenter may wish to study the effect of controllable large environmental differences i.e. macro environmental differences on genotypes more in the nature of "treatments". For instance, he may like to study the effects of differences in locality, or food supply or season or different breeds or different genetic strains. If a specific change in the environment does not have the same effect on different genotypes, it is called in statistical sense "genotype and environmental" interaction. The interaction may be in several forms. For instance, a specific difference in environment may have a greater change on some genotypes than on others or the change may be in opposite direction or the order of merit of different genotypes is not the same in different environments. In other words, the existence of genotype-environments interaction may mean that the best genotype in one environment is not the best in another environment. For instance the best dairy breed in temperate climate may not be the best breed in tropics. Note that for illustrating genotype-environment interaction we have used breeds or strains which are not homozygous lines. A breed or a strain is a mixture of genotypes or in other words they are genetically heterogenous. But different breeds or strains are different mixtures of genotypes and they differ in their mean genotypic values and we are considering how these mean values behave in different environments.

As an example of interaction between genotypes within a breed and environments we shall consider the results of progeny tests reported by Prichner (1969) for four bulls which were tested simultaneously in experimental stations and in field farms. The following are the average milk yield of large number of daughters born to bulls noted against each bull under the two environments.

	Results of	
Bull	Station test	Field test
A	4380	3760
B	4350	3560
C	4130	3880
D	4060	3640

We find that the milk yield is higher at experimental stations compared to field units possibly due to good feeding and management conditions in experimental stations. There is also genotype environmental interaction. Bull A was the best when station tested and bull C was the best when field tested. This is important to the animal breeder since it implies that station tested bulls may not be superior when used in field.

EXERCISE

1. What is meant by variance? Give its components with symbols.
2. How the variance can be measured?
3. Give an account on the estimation of genotypic frequency.
4. Describe on the genotype and environment interaction.
5. The variances of leaf number in the F_1 and F_2 generations of a cross of tobacco varieties were 1.46 and 5.97 respectively. Estimate the degree of genetic determination in the F_2 generation.
6. The variance of leaf number in the F_1 and F_2 generations of a cross of tobacco varieties were 3.14 and 7.86 respectively. Estimate the degree of genetic determination. What are the assumptions to be made to estimate the degree of genetic determination in F_2?

Chapter 30

Repeatability

In certain conditions, more than one measurement of a character may be made on an individual. Examples are lactation yield in successive parity, litter size in successive pregnancies, wool yield in different years. Hence there are variations between individuals and within individual between measurements of the same character. The between individuals component is partly environmental and partly genetic. The environmental part is due to permanent environmental variation. The within individual component is entirely environmental in origin caused by temporary differences of environment between successive performances i.e. between measurements. Then between animal variance is due to permanent differences between animals which arise from the differences in the genotype between individuals and from differences in permanent environmental factors arising early in ~~life~~ and causing a permanent effect on the animal. Hence the phenotypic variance may be written as

$$V_P = V_G + V_{EP} + V_{ET}$$

Where V_{EP} and V_{ET} represent permanent and temporary components of environments. The between individuals environment variance is also known as general and within individuals component as special environmental variances. The ratio of the between individual component to the total phenotypic variance is the intra- class correlation. It is the correlation between repeated measurements on the same individual and is known as repeatability of the character. It is the correlation of the successive performances of the individual.

So repeatability

$$R = \frac{V_G + V_{EG}}{V_P}$$

Thus the repeatability expresses the proportion of the variance of single measurements that is due to permanent differences between individuals.

Estimation of repeatability:

It is estimated by obtaining the correlation between records of the same animal. When only two records are involved, straight intraclass correlation can be calculated, when more than two records are involved estimate can be obtained by intraclass correlation by Analysis of variance.

Analysis of variance of multiple measurement

Source of variation	Degrees of freedom	Sum of squares	Mean squares
Between animals	(S-1)	$\sum \frac{X1.2}{n1} - \frac{X2}{N} = SSB$	$\frac{SSB}{S\text{-}1} = MSB$
Within animals	(n-s)	$\Sigma i \Sigma j \; X^2 ij \; - \frac{X12}{ni} = SSW$	$\frac{SSW}{n-s} =$ MSw

Where S = number of animals
ni = number of records for within animal
n = total number of records

Between animal component of

$$\text{variance } \sigma^2 B = \frac{MSB - MSW}{ro}$$

Where ro is the number of records per animal, if all the animals have the same number of records. If the animals differ in the number of records, the ro is estimated by

$$ro = \frac{1}{(s\text{-}1)} \quad n - \frac{(ni2)}{n}$$

The within animal component of variance $\sigma^2 W$ = MSW

Intraclass correlation = Repeatability

$$\text{Repeatability} = R = \frac{\sigma^2 B}{\sigma^2 B + \sigma^2 W} \quad \frac{V_g + V_{EP}}{V_P}$$

Characteristics of Repeatability:

It measures the correlation between the repeated measurements of the same individual. It indicates the gain in accuracy that may be expected from the use of the mean multiple measurements instead of single measurement.

For example milk production is repeated through many lactations. Similarly egg production is repeated for 3 to 4 years and would yield twice in a year for 2 to 3 years. When the repeatability is high, there may be very little variance due to temporary environmental changes, multiple measurements may lead to little gain in accuracy. When the repeatability is low, there may be considerable temporary environmental variance and reduction of them may lead to worthwhile increase in accuracy. Repeatability value will be ranging from 0 to 1. It is essentially an upper limit for heritability. It is due to both the genetic constitution of the individual and environmental factors. But heritability is due to genetic constitution alone. Hence repeatability will always be more than heritability.

Uses of repeatability:

It sets the upper limit of the degree of genetic determination. It shows how much is to be gained by the repetition of measurements and predicts the future performance from the past records. And it enlightens the nature of environmental variance.

The knowledge of repeatability is necessary for predicting the lifetime production of an animal. If it is very high, the animal can be kept or culled based on the first record of observation. On the other hand if the repeatability is low, more than one observation on the same character is necessary for a decision on the animal. If you wish to select animals taking their previous performance as a guide to improve their future performance in the current herd, the following equation gives an estimation of their real producing ability.

$$\text{MPPA} = M + bR\,(x_i - -M)$$

Where

MPPA = Most probable producing ability

$$b = \frac{n}{1 + (n-1)R} \text{ where}$$

n = no. of records

R = Repeatability of the trait

Xi = mean of the ni records of the ith individual

M = Population mean

Thus for each animal from the mean of its records, the MPPA may be estimated and the individuals with the best estimates are chosen in order to improve the subsequent production level in the herd.

EXERCISE

1. Define repeatability and discuss on its properties and uses.
2. How will you estimate repeatability?
3. A sample of 10 rabbits have the following number of young ones in their first and second pregnancies:

Rabbits	1	2	3	4	5	6	7	8	9	10
Pregnancy I	11	9	13	10	9	8	10	11	10	13
Pregnancy II	10	12	12	10	8	6	12	9	12	12

Estimate the repeatability of litter size in rabbits.

4. The following are the annual wool yield from 6 Merino x Nilgiri cross bred sheep for 5 consecutive years, at Sheep Breeding Research Station, Sandynallah. Estimate the repeatability of wool yield.

	Sheep					
Years	S_1	S_2	S_3	S_4	S_5	S_6
1	1.2	0.9	1.4	1.1	0.8	1.2
2	1.2	1.2	1.3	1.2	0.9	1.1
3	1.5	1.3	1.5	1.5	1.2	1.3
4	1.6	1.2	1.6	1.7	1.1	1.2
5	1.5	1.4	1.5	1.6	1.3	1.5
	7.0	6.0	7.3	7.1	5.3	6.3

5. The following sizes in the I and II litters of 10 sows are given below. Find the repeatability of litter size in pigs.

Sows	1	2	3	4	5	6	7	8	9	10
Litter I	7	6	7	5	7	8	9	9	5	11
Litter II	9	5	6	10	9	8	8	7	6	9
Total	16	11	13	15	16	16	17	16	11	20

Chapter 31

Heritability

We can obtain the phenotypic values by measuring the individuals directly. It is the breeding value that influence the phenotypic value of the next generation. If we select individuals based on their phenotypic values, success in improvement can be predicted only from the knowledge of correspondence between phenotypic value and breeding values. This degree of correspondence is measured by heritability. The degree of genetic determination VG/VP though may measure the relationship between phenotype and genotype, it is of little practical use, since to estimate genetic determination highly inbred lines are required which may not be available in farm animals. Since the genotypic values are determined by the additive effects of the genes, heritability estimation is necessary to obtain the best estimate of an individual's breeding value.

Genes are the units inherited by the individuals from their parents. Because the genes are transmitted independent of their alleles, and of the genes at other loci, only additive effects i.e. breeding values of the parents will determine the genotypic values of their progeny.

Heritability is the ratio of additive genetic variance to the total phenotypic variance.

$$\text{i.e. } h^2 = \frac{V_A}{V_P}$$

It expresses the proportion of the total phenotypic variance that is attributable to the average effects of genes. In statistical sense it is the regression of additive gene effect (breeding value) on the phenotype

$$h^2 = b_{AP}$$

The customary symbol h^2 derives from Wright's (1921) terminology.

Regarding the heritability as the regression of breeding value on phenotypic values, we see that the best estimates of an individuals breeding value is the product of its phenotypic value and heritability of the trait measured.

$$E(B.V) = h^2P$$

Because of its predictive role in estimating the breeding value of an individual the heritability finds its application in almost all the equations used for predicting the progress in selection experiments. Therefore estimation of heritability is of primary interest in genetic studies.

Salient features of heritability:

1. It is ranging from 0 to 1.
2. h^2 of metric traits is one of the important properties of that trait.
3. h^2 is not only the property of the character but also of the population of the individuals.
4. h^2 of a character differs from population to population and from time to time.
5. The value of h^2 depends on the magnitude of all components of variance and hence a change in any one of the components will affect the h^2.

6. If h^2 is high, selection will be effective and if it is low, selection will be ineffective.
7. All reproductive traits will be low heritable.
8. If all the phenotypic variance is due to genetic effect, then

$$V_P = V_A \; (V_E=o)$$

$$h^2 = 1$$

If all the phenotypic variance is environmental in nature

$$\text{i.e. } V_P = V_E \; (V_A = 0)$$

$$\text{then } h^2 = 0$$

If the half of the phenotypic variance is due to gene effects.

$$\text{i.e.} = V_A = 1/2V_P$$

$$h^2 = 0.5$$

If the environmental components of variance is three times as large as genetic components.

$$\text{i.e. } V_E = 3V_A$$

$$h^2 = \frac{V_A}{V_P} = \frac{V_A}{V_A+V_E} = \frac{V_A}{V_A+3V_A} = \frac{1}{4}$$

$$h^2 = \frac{1}{4}$$

9. In the broad sense h^2 involves all types of gene action. In complete dominance, when a gamete carrying the active dominant allele A unites with a gamete bearing the recessive allele a, the resulting phenotype might be two units. When two A gametes unite, the phenotypic result would be still two units. But if the genes lacking dominance i.e. additive genes are involved then the A gamete will add one unit to the phenotype of the resulting zygote regardless of the allelic contribution of the gametes with which it unites. Thus only the additive genetic component of variance has the quality of predicting the results in the

breeding plans. Heritability in the narrower sense is the ratio of additive genetic variance to the total phenotypic variance.

10. The h^2 of some metric traits:

Cattle: Milk yield - 0.2 - 0.4
Fat percentage -- 0.4 - 0.8
Calving interval - 0.15
Sheep: Wool yield - 0.3 - 0.6
Body weight - 0.2 - 0.4
Poultry: Egg yield - 0.2 - 0.4
Egg weight - 0.3 - 0.4
Pig: Litter size - 0.1 - 0.2

Uses of heritability:

It expresses the reliability of the phenotypic value as a guide to the breeding value.

It tells the degree of correspondence between the phenotypic and breeding values that determines the success of selection.

It is the best estimate of individuals breeding value, hence it finds its application in all equations used for predicting the speed of progress in selection experiments. Hence estimation of heritability is of primary interest in all genetic studies.

For precise genetic analysis estimation of h^2 is essential and hence it is included in all formulas of breeding experiments.

Estimation of heritability:

To measure heritability it is necessary to partition the phenotypic variance into additive versus the rest. The relatives resemble each other because they possess genes identical by descent and the identical genes being identical in state cause the same effect in both the individuals. Therefore a measure of the degree of resemblance will provide an estimate of additive variance in the population. The most commonly encountered relationships are between offsprings and their parents and between full sibs or half sibs. The degree of resemblance between offspring and parents is measured by regression coefficient and that between full or half sibs is measured by correlation coefficient.

Let a population be made up of several families of full or half sibs. By analysis of variance we can partition the total observed variance into between and within families. The within family variance is the variance of the individuals about their family means and the between family variance is the variance of the 'true' means of the families about the population mean. The variance of within families equals the covariance expected between two randomly chosen individuals of a family. Similarly variance between families of full or half sibs is equivalent to covariance between full or half sibs. The resemblance between related individuals can be looked as either similarity of individuals within families or as difference between individuals in different families. The greater the similarity within families, the larger will be the difference between families. The degree of resemblance can therefore be estimated as the ratio of between family component to the total phenotypic variance which is called intra-class correlation.

$$t = \frac{\sigma^2_B}{\sigma^2_B + \sigma^2_W}$$

The offspring-parent resemblance can more easily be estimated by regression coefficient.

$$b_{op} = \frac{Cov_{OP}}{\sigma^2_P}$$

where Cov_{OP} = covariance of offspring and parents and 6^2_P is the variance of the parents.

Therefore four methods are in practice for estimating the heritability.

I Regression method
- Offspring and one parent
- Offspring and mid parent

II Intraclass correlation
- Full sibs
- Half sibs

Environmental covariance between relatives:

Relatives resemble each other not only due to identical genes they possess but also from the similarity of environments in which they live. For instance, full sibs, particularly if they belong to the same litter or clutch such as litters of pig or mice, are contemporary, enjoy the same maternal, housing, season and other environmental effects. This component of environment which contributes to similarity of members within families gives rise to differences between families. Thus the variance, consequently covariance between families are increased. We may denote this between group environmental component as variance due to "common environment", V_{EC} and the remainder of the environmental variance which appears in 'within' group component as variance within group V_{EW}.

Differences in the similarity of environments between unrelated individuals led to the partition of the total variance into "variance due to common environment" and "variance within".

i.e., $V_E = V_{EC} + V_{EW}$

Since in the presence of V_{EC}, the observed covariance between relatives would be more than what would be obtained from out of genetic covariance alone, the V_{EC} has to be regarded as a complication and need to be eliminated. Common source of V_{EC} in mammals is maternal effect. Falconer (1960), reported that in mice large mothers tend to provide better nutrition to their offspring both before and after birth than small mothers. Therefore young of large mothers tend to grow faster and thus mother and its offspring tend to resemble each other in body size. Covariance between full sibs in general include large amount of V_{EC}. Juvenile traits, in general show much more variance due to differences in maternal effects than adult traits. Since V_{EC} may be mainly due to differences in maternal influence, it is sometimes

designated as V_M.

Phenoptypic covariance between relatives : The sum of genetic and environmental covariance between relatives yields the phenotypic covariance between relatives.

Relatives	Covariance*
Offspring and one parent	$Cov_{OP} = 1/2V_{(A)}$
Offspring and mid parent	$Cov_{OP} = 1/2V_{(A)}$
Half sibs	$Cov_{HS} = 1/2V_{(A)}$
Full sibs	$Cov_{FS} = 1/2V_{(A)} + 1/4V_D + V_{EC}$

* Interaction components are omitted since they are generally negligible.

Note that Cov_{FS} may contain non-additive genetic (V_D) and maternal variance (V_{EC}) in addition to additive variance if they are important in the control of the trait.

Offspring and one parent:

The degree of resemblance between offspring and parent as already pointed out is measured by regression.

$$b_{op} = \frac{\text{Covariance offspring - parent}}{\text{variance of parent}}$$

The covariance is that ratio of the genotypic value of individual to the mean genotypic values of the offspring produced by mating at random in a population. The mean value of the offspring is by definition is half the breeding value of the parent. Therefore the covariance is that ratio of an individual's genotypic value with half of its breeding value i.e.

$$Cov_G \text{ X } 1/2A$$

Multiplying this totgether, the covariance of offspring and parent will be

$$Cov_{op} = 1/2\,V_A$$

and the variance of parent is the phenotypic variance V_P

$$\text{Therefore bop} = \frac{1/2V_A}{V_P} = \frac{1}{2}h^2$$

Offspring and mid parent:

If the P and P' are the values of the parents, then the

mid parent value = 1/2 (P+P')

$$\text{then } Cov_{op} = \frac{\text{Covariance offspring — Mid parent}}{\text{variance of parent}}$$

If both parents have the same variance, then

$$Cov_{op} = CoV_{op'}$$

$$CoV_{op} = 1/2V_A$$

If the variance in the two sexes are equal variance of male parent V_m = variance of female parent V_F = Phenotypic variance V_P

Variance of mid parent value = V_{ar} {(1/2(M+F)}

$$= \frac{Cov.Op}{V_P} = 1/4\ Var\ (M+F)\_$$

—

$$= 1/4\ \{(Var(M) + Var\ (F)\}$$

$$= 1/4\ (2V_P) = 1/2\ V_P$$

Therefore bop = $1/2\ V_A / 1/2V_P$

$$= \frac{V_A}{V_P} = h^2$$

Half sib analysis:

Half sibs are individuals which have one parent in common. Thus the mean genotypic value of the group of half sibs is by definition half the breeding value of the common parent. Half sib analysis consists of choosing a group of sires at random mating each of them to several dams and

measuring one progeny per dam. The offspring of the same sire by different dams constitute half sibs. This experimental design is useful for uniparous animals like cattle.

Assume that there are S sires, each mated to several dams and producing one progeny per dam. The observation on the jth offspring of the ith sire may be considered to have the

$$Y_{ij} = \mu + s_i + e_{ij}$$

where u = popualtion mean, s_i = ith sire effect and eij = random error. All the effects except u are assumed as random. The analysis may be carried out using the conventional procedure for the analysis of one-way lay out. The expected mean squares are tabulated in table.

Analysis of variance of half sib design

Source of variation mean	Degrees of freedom	Mean of squares	Expected squares
Between families	S - 1	MS_S	$6^2_w + n6^2_s$
Within families	n - s	MS_W	6^2_w

where S = the no. of sires; n_i = number of offspring for ith sire. n = the no. of offspring per sire if equal subclass number exists or else, the average no. of offspring per sire (r_o) may be determined by equation.

$$\text{Interclass correlation is } t = \frac{\sigma^2_\sigma}{\sigma^2_\sigma + \sigma^2_\omega}$$

The estimate of heritability is

$$h^2_{HS} = \frac{4\sigma_s{}^2}{\sigma S^2 + \sigma^2 W}$$

$$t = \frac{4\sigma_B^2}{\sigma S^2 + \sigma^2_W} = \frac{CoV_{HS}}{V_P}$$

$$= 1/4\ V_A/V_P = 1/4\ h^2$$

Estimation of h^2 by sib correlation may be less precise than regression method if the heritability is more than 0.25. However, correlation method for estimating h^2 requires data of only one generation while regression method may need data of two consecutive generations, parental and offspring generations.

Full sib analysis:

Full sibs have both parents in common.

In multiparous animals with larger full sib families like pigs and poultry, full sibs can be used for estimating heritability. A simple experimental design will be to mate each sire with one dam and to measure n offsprings per mating. The members of each family are full sibs and the same analysis of one-way lay out can be used and the correlation t is interpreted as the correlation between full sibs. The heritability is

$$h^2_{FS} = 2\ t$$

if it can be assumed that there is no epistasis, no dominance and no environmental correlation, all of which are non-realistic. More often the full sib covariance is augmented by the presence of environmental correlation between litter mates caused by maternal effects.

The mean genotypic value of the full sib is equal to the mean breeding value of the parents.

Let A and A' are the breeding value of the two parents, then

$$COV_{FS} = 1/2\ (A+A')$$

$$= 1/4\ (V_A + V_A) = 1/2\ V_A$$

Let the parents have the genotypes A_1A_2 and A_3A_4, then there will be 4 genotypes among the progeny, i.e. A_1A_3, A_1A_4, A_2A_3, A_2A_4, each with a frequency of 1/4. The first sib will have any of these genotypes. The probability that the second sib chosen will have 1/4 frequency. 1/4 of all sib pairs will have the same genotypes and consequently have the dominant deviation 'D'. The mean cross products of the sibs will be $1/4V_D$, then $COV_{FS} = 1/2\ V_A + 1/4V_D$.

The intraclass correlation between full sibs.

$$t_{FS} = \frac{COV_{FS}}{V_P}$$

$$= \frac{1/2\ V_A + 1/4\ V_D + V_E}{V_P}$$

$$= > 1/2\ h^2$$

Full sibs can be used for estimating h^2 in multiparous animals with large number of full sib families like pigs and poultry. A more common form in which data are being obtained from multiparous animals is that by mating each sire to several dams and measuring number of offspring per dam. In reality the sub class numbers are unequal. Let the number of sires be S, each of them mated to different number of dams and each dam produces different number of offsprings. Let D be the total number of dams and n_{ij} is the number of progenies for j^{th} dam mated to i^{th} sire.

Analysis of variance

Source of variation	Degrees of freedom	Sum of squares	Mean squares
Between sires	S-1	SS_s	MS_s
Between dams within sires	D-S	SS_D	MS_D
Offspring within dam & sires	n-D	SS_W	MS_w

The components can be estimated by equating the observed mean squares to their expectations.

$$\sigma^2 w = MS_w$$

$$\sigma^2 D = \frac{MS_D - MS_w}{n}$$

$$\sigma^2 S = \frac{MSs - MSD}{nD}$$

h^2 from sire component of variance $h^2S = \dfrac{4\ \sigma^2 S}{\sigma^2 S + \sigma^2 D + \sigma^2 W}$

h^2 from the dam component of variance

$$h^2 D = \frac{4\sigma^2 D}{\sigma^2 s + \sigma^2 D + \sigma^2 w}$$

h^2 from sire + dam components of variance

$$h^2\ S{+}D = \frac{2(\sigma^2 S + \sigma^2 D)}{\sigma^2 s + \sigma^2 D + \sigma^2 w}$$

h^2 is estimated by the regression or correlation methods. To estimate h^2 through the resemblance between half sibs, it is only necessary to compute intraclass correlation and multiplied by 4 or to compute the regression coefficient of offspring on mid parent which itself estimates the heritability.

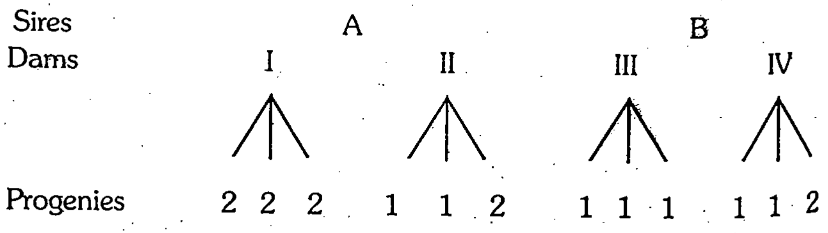
Choice of method for estimating heritability:

- Absence of non-additive genetic causes of resemblance in a relationship is the most important criterion in choosing a method for determining heritability. The genetic cause of resemblance between full sibs is not confined to the additive variance and the correlation is augmented by part of the non-additive genetic variance (V_D) and maternal variance (V_{EC}) if any present. Therefore an estimate of the heritability from full sibs is, strictly speaking, valid only in the absence of non-additive genetic and maternal variance. Maternal variance is also likely to augment the covariance between offspring and dam. For these reasons the correlation between full sibs and sometimes the regression of offspring on dam are avoided as means of estimating the heritability.

The choice of method is also a matter of deciding which gives the most precise estimate for the same total of animals measured with the given facility. In general, low heritability are more efficiently estimated from the half sib correlation and heritabilities higher than about 0.25 are more efficiently estimated by offspring-parent regressions. However, one difficulty with offspring-parent regression method is that we need data from two consecutive generations.

EXERCISE

1. Give definitions of heritability.
2. Give an account on its characteristics and its uses.
3. Describe in detail on the different methods of estimating heritability.
4. Write short notes on
   a) Environmental covariance between relatives.
   b) Phenotypic covariance between relatives.
5. Write in detail on the half sib analysis of heritability and its merits.
6. Discuss on the full sib method of estimating the heritability and its merits and demerits.
7. Which methods are better methods for estimating the heritability? Give reasons for it.
8. The total genetic variance of 180 day body weight in a population of swine is 100 kg^2. The variance due to dominance effect is 30 kg^2. The variance due to epistatic effects is 10 kg^2. The environmental variance is 200 kg^2. What is the heritability estimate of this trait?
9. Determine the a) dominance variance and b) environmental variance from the following information.
   Heritability = 0.3 —
   Phenotypic variance = 100 kg^2, total genetic Variance = 60kg$^{2'}$ and epistatic variance is absent
10. Two sires each mated to two dams and three offsprings per dam measured for wool yield in kgs as presented below.



Estimate the significance of between sires and between dam variance. Estimate the causal components of variation and represent in terms of percentage.

Determine h^2 of the trait of wool yield through sire and dam components.

11. The following are the first lactation milk yield on 100 kilo units of 20 cows which are the progenies of 5 sires. Estimate the h^2 by half sib correlation method.

Sire No.	Dam No.	Daughter's yield	Total of sires
S_1	1	22	95
	2	18	
	3	30	
	4	25	
S_2	5	30	78
	6	20	
	7	16	
	8	12	
S_3	9	16	60
	10	17	
	11	12	
	12	15	
S4	13	16	80
	14	22	
	15	24	
	16	18	
S5	17	17	77
	18	20	
	19	21	
	20	19	
			390

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Chapter 32

Correlation Among Traits

A character is described by numbers, mean variance, standard deviation in a population. While considering two traits covariance is considered which is the simultaneous variation among the traits. Correlation is based on covariance. To study correlation minimum two traits are required.

Two characters may be correlated to the extent that both are influenced by the same genes and by common environmental factors. The correlation arising from genetic cause is called as genetic correlation. The correlation arising from common environmental cause is known as environmental correlation.

The observed correlation arising from the combined effects of genotypes and environment is known as phenotype correlation.

Phenotypic correlation:

A correlation may also be defined as the ratio of the appropriate covariance to the product of the two standard deviations.

The phenotypic correlation between two traits 1 and 2 is

$$r_p = \frac{Cov(P_1P_2)}{\sigma P_1 . \sigma P_2}$$

$Cov(P_1\ P_2)$ is the phenotypic covariance between 1 and 2 traits.

σP_1 and σP_2 are the phenotypic standard deviations of 1 and 2 traits.

If both characters have a high h^2 the phenotypic correlation is almost, entirely genetic and in low h^2 characters the phenotypic correlation is entirely environmental.

The genetic and environmental correlation may differ in magnitude and sometimes in sign also. A difference in sign between the two correlations indicate that the genetic and environment sources affect the characters through different physiological mechanisms.

Example in poultry:

		r_p	r_G
18 weeks body wt.	Egg production upto 72 weeks of age	0.09	-0.16

r_G and r_E differ in sign. The pullets that are heavier at 18 weeks due to genetic reasons reach sexual maturity later and lay few eggs. Pullets heavier due to environmental causes matures earlier and lay more number of eggs.

Genetic correlation:

The chief cause of genetic correlation is pleiotrophy. A gene is said to be pleiotyophic in effect if it affects many traits. Pleiotrophy is a common property of major gene. For eg. the genes that promote growth may influence the trait by increasing the height and weight of the individuals. These traits are correlated due to pleiotrophic effect of genes.

$$r_G = \frac{Cov\ (A_1\ A_2)}{\sigma A_1\ \ \sigma A_2}$$

Where Cov $(A_1\ A_2)$ is the covariance of additive genetic effects of 1 & 2 and σA_1 & σA_2 are the additive genetic standard deviations of 1 & 2 traits.

Uses of genetic correlation:

Selection for a character affects favourably or adversely the other character when two characters are correlated, selection for one trait will bring about change in other trait. This response is through correlated response. For eg. broiler wt. and feed efficiency (feed cons/wt) are negatively correlated. Hence selection for increasing broiler weight will improve the feed efficiency also. Body weight can be easily measured while feed efficiency is difficult to measure. Hence improvement in feed efficiency can be brought through correlated response by selection for body weight.

Egg production & egg size	negative correlation
Milk yield & fat %	negative correlation

Hence a knowledge of genetic correlation of traits will be useful to forecast reduction in the correlated traits due to single trait selection.

Environmental correlation:

Environmental covariance and variance are obtained by substracting additive genetic covariance and variance from the phenotypic covariance and variance.

$$= \frac{COV\ (E_1\ E_2)}{\sigma E_1\ \ \sigma E_2}$$

Where Cov $E_1.E_2$ is the environmental covariance. σE_1 σE_2 are the standard deviations of the environmental values of 1 and 2 traits.

Examples for correlation among economic traits:

	r_p	r_A	r_E
Milk yield: Fat %	-0.26	0.38	-0.18
Milk yield in 1st, 2nd lact.	0.40	0.75	0.26
Poultry:			
Body weight: egg weight	0.33	0.42	0.23
Body weight: egg production	0.01	-0.17	0.08
Egg weight: egg production	-0.05	-0.31	0.02

Characteristics of correlation:

Correlation among traits may be positive or negative or no correlation. Positive correlation is the one in which the improvement in one trait may improve the other trait also, or both traits may show decline. When increase in one character brings decline in another trait it is negative correlation. When the effect of one trait is in no way influencing the other trait, then both are not correlated and they are independent.

Positive correlation	**Negative correlation**
Feed efficiency & growth	Milk yield & fat percentage
age and height	

No correlation

Horn length and milk yield

Correlation coefficient is the measure of correlation between traits

It ranges from 0-1

0 - No correlation - The traits are independent.

1 - Perfectly correlated.

When the characters are related among themselves due to genetic constitution, selection becomes very effective. While selecting for one character simultaneous improvement is possible in other character also. In selection programmes, therefore it is desirable to obtain estimates of genetic correlation.

Rate of gain and milk yield are negatively correlated. Therefore an attempt to produce a high milk producing and meat producing breed of cattle will be a failure. Therefore it will be most desirable to develop separate kinds for milk and beef.

Similarly milk production and work are negatively correlated. Hence our attempts to produce Indian cattle with high milk production and high draught power have not succeeded.

EXERCISE

1. What is the basis of correlation. Classify the correlation among traits and discuss on it.
2. Write briefly on phenotypic correlation.
3. Discuss on the characteristics and uses of genetic correlation.
4. Following are the records of respiratory rate of male calves of less than a year in age and observations on air temperature and humidity recorded at the times of taking the respiratory rate. Obtain the correlation—

a) between respiration and air temperature after eliminating influence of humidity,

b) between respiration and humidity after eliminating the influence of air temperature.

Sl.No.	Respiration rate per minute	Air temeprature	Humidity
1.	16	77	59
2.	22	85	45
3.	20	75	75
4.	24	84	49
5.	22	76	70
6.	23	83	50
7.	24	84	85
8.	17	74	77
9.	18	73	68
10.	27	88	35
11.	18	75	73
12.	25	85	45

Chapter 33

Response to Selection

The basic effect of selection is to change the gene frequencies. The effects of selection can be described in terms of the observable properties - mean, variances and covariances eventhough the underlying cause of changes is the change of gene frequencies. To describe the change of the genetic properties from one generation to the next we have to compare successive generations at some point in the life cycle of the individuals, and this point is fixed by the age at which the characters under study is measured.

There are 2 types of selection

1) Artificial 2) Natural

In artificial selection the breeder chooses the parents of the next generation. The natural selection operates through differences in fertility among the parents and differences in viability among the offsprings.

The basic effect of selection is to change the gene frequencies of the population. The genes favouring the traits will get accumulated at all loci controlling the trait. Genetically selection act in two ways. 1) changing of gene frequencies 2) Disturbing the H.W. equilibrium.

Though selection is relaxed, the change in gene frequency will remain permanent but H.W. equilibrium will be restored.

Therefore there are three stages at which a change of gene frequency may result due to selection.

1) Through artificial selection among the adults of parent generation.

2) Through natural differences of fertility also among the adults of the parents generation.

3) Through natural differences of viability among the individuals of the offspring generation.

Response to selection:

For traits influenced by many genes, the genotypes and also the phenotypes have distributions approaching a normal curve. If superior individuals are selected, those genotypes possessing more plus genes are chosen. Only selected individuals reproduce. Therefore the progeny generation will have a higher frequency of plus genes and the progeny average performance will be higher than the average performance of the parental generation before selection. The change produced is the response to selection which we shall symbolise by R; it is the difference of mean phenotypic value between the offspring of the selected parents and the whole of the parental generation before selection. The measure of the selection applied is the average superiority of the selected parents, which is called the selection differential and will be symbolised by SD. It is the mean phenotypic value of the individuals selected as parents expressed as a deviation from the population mean, that is from the mean phenotypic value of the individuals in the parental generation before selection was made.

The regression of the additive genotype (breeding value) on phenotype is the heritability coefficient. Regression refers to the increase expected in the breeding value when the phenotype increases by one unit. Thus response is given by $R = h^2 SD$ which provides a means of prediction based on observation made only on the individuals of the parent generation before selection. The prediction of response is valid in principle for only one

generation of selection since the heritability of the character will be changed as the gene frequencies are altered due to selection.

Selection differential (SD)

The response depends on the heritability of the character and on the amount of selection applied as measured by the selection differential. The magnitude of the selection differential depends on two factors: the proportion of the population included among the selected group and the phenotypic standard deviation of the character. The dependence of the selection differential on these two factors is illustrated diagrammatically. The individuals with highest values are supposed to be selected so that the distribution is sharply divided at a point of truncation, all individuals above this value being selected and all below rejected.

The arrow in each figure marks the mean value of the selected group, and S is the selection differential. In graph (a) half the population is selected and the selection differential is rather small; in graph (b) only 20 per cent of the population is selected and the selection differential is much larger. In graph (c) 20 per cent is again selected but the character represented is less variable and the selection differential is consequently smaller. The standard deviation in (c) is half as great as in (b) and the selection differential is also half as great.

The proportion selected may differ in two sexes. In animal breeding normally few males than females are selected.

To maintain constant population size, in cattle about 2/3 of female calves are retained while only 5% of bull calves are needed for replacement under natural service. In A.I, only 1% is needed. In pigs and poultry 10-15% of female and 1-3% males needed only be selected.

Hence selection applied to males and females differ, the value of S are unweighed means for the two sexes.

$S = 1/2\ (Ss + S_D)$

If the trait is sex limited such as milk yield, selection is done only in females.

$S = 1/2\ S_D$

Even for the same proportion selected the selection differential will vary if the population differ in their standard deviation. This problem can be overcome by expressing the selection differential as standard deviation unit.

Intensity of selection:

SD can be expressed as $\frac{S}{\sigma P}$

This standardised selection differential is called as the intensity of selection which is symbolised as i.

The intensity of selection measures, how many standard deviation by which the mean of the selected population differs from the mean of the population from which they were selected.

$i = S/\sigma P.$

$S/\sigma P$ is a generalised measure of selection differential by which we can compare different methods of selection.

Generalised response:

The response to selection can also be estimated by the formula.

$R = h^2S$

This can be rewritten

$$R/\sigma P = \frac{h^2S}{\sigma P}$$

$R/\sigma P$ is a generalised response. By this we can compare the response for different traits or for the same trait in different populations.

$R/\sigma P = h^2\, S/\sigma P$

$R/\sigma P = ih^2 \qquad i = S/\sigma p$

$R = ih^2\, \sigma P$

Since $h^2 = \sigma A/\sigma P$

$$R = ih\frac{\sigma A}{\sigma P} = ih\,\sigma A$$

h is the correlation between phenotype and B.V. of the trait hence h = rAP

Then $R = irAP\sigma A.$

This formula is used for comparing the different methods of selection.

Prediction of response:

The response to selection can be estimated by this formula

$R = h^2S$

Proof:

From the mean phenotypic value of the parents, the mean phenotypic value of the offspring can be predicted from the regression equation

$E\,(Xo/XS/D) = a + bs \times S + bD \times D$

Where Xo/Xs X D is the mean phenotypic value of the offsprings from to sire S and dam D, with the phenotypic values of XS XD respectively bs and b_D are regression of offspring on sire and dam respectively.

Let Ss & SD be the selection differential for the sires and dams selected.

$$\begin{aligned} R &= bs\,Ss + b_D\,S_D \\ &= b_{op}\,Ss + b_{op}\,S_D \\ &= b_{op}\,(Ss + S_D) \\ &= b_{op}\,s \\ \because\ h^2 &= b_{op} \\ R &= h^2S \end{aligned}$$

Falconer gave a simple proof using genetic theory. The deviation of the progeny from population mean is by definition, the breeding value of the parents i.e.

$$\begin{aligned} R &= \text{BV of parents} \\ &= h^2S \end{aligned}$$

The effect of selection is to change the gene frequencies. Consequently h^2 of the trait selected may be changed. But the change in gene frequency will take place after 5 to 10 generations only. Hence h^2 need not be redetermined for each generation.

Rate of Response:

The rate of response is given by the equation

R = irAPσA

This shows that the magnitude of response depends on 3 factors.

i = intensity of selection

rAP = correlation between breeding value and selection criterion

σA = Genetic variability

Even if any one of the above is O, then response will also be O.

How to improve this:

A breeder has no control over the genetic variability for the trait (σA) except perhaps by improving the h^2 by reducing environmental variation through attention to techniques of rearing and management.

Hence there are only two ways open for the breeder to improving the response.

1) by increasing the correlation between the breeding value and selection criteria

2) by increasing the intensity of selection.

Generation Interval:

$R = h^2S$ gives the response per generation. But the genetic progress per year will be of more practical significance. This can be obtained by dividing the equations with generation interval.

$$\frac{R}{1} = \frac{h^2S}{1} = \frac{ih^2\,\sigma P}{1} = \frac{ih\,\sigma A}{1}$$

The generation interval is the average age of the parent when their offspring horn.

Realised h^2:

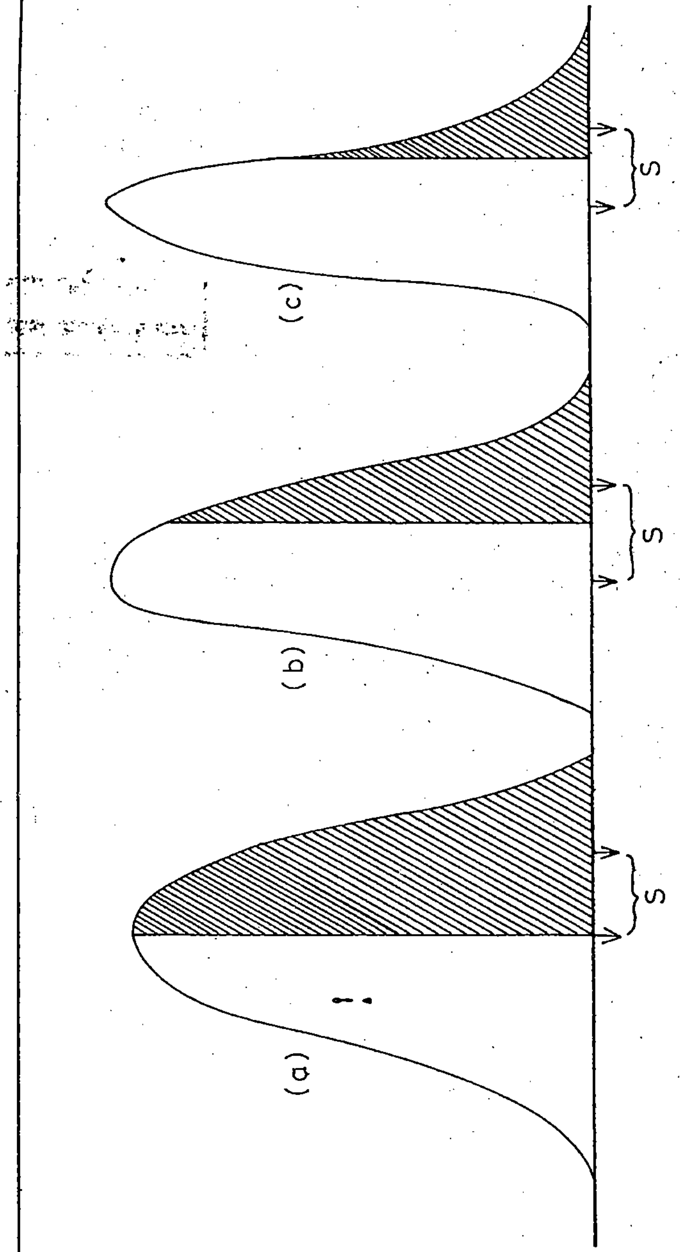
The equation of response $R = h^2S$ showed the relation between the response and S.D. By rearranging the equation we can find that h^2 can be estimated.

$$h^2 = R/S$$

The h^2 estimated by this way is called the realised h^2. This can be estimated as a regression of response on cumulated selection differential in selection or breeding programmes continued over a few generations.

Correlated response:

When two characters are correlated selection for one trait will bring about change in other trait. The expected response of a character Y when selection is applied to another character X may be deduced. Milk yield and fat percentage in dairy cattle are negatively correlated improvement in milk yield may result in decrease in fat percentage. This kind of response is one character due to selection in other character is termed as correlated response.



Selection Differential

All the individuals in the stippled areas beyond the points of truncation are selected
a) 50% selected
b) only 20% selected
c) Only 20% selected with standard deviation is half as great as (a). The character is less variable

EXERCISE

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पुस्तक खराब करु नये.

1. What are the three stages at which the gene frequency is altered?
2. Define selection differential and discuss on it.
3. Give in detail on response to selection.
4. Write short answers on
    a) Intensity of selection
    b) Generalised response
    c) Prediction of response
    d) Generation interval
    e) Realised heritability
    f) Correlated response
5. What is the rate of response and how to improve it?
6. The average broiler weight at 8 weeks age in a group of birds with equal number of male and female were 1200 and 1000 gm. 10 cockerals and pullets selected from their group weighed on a average 500-1100 gm respectively. The h^2 of body wt. at 8 weeks of age was 0.45. Estimate the expected response and expected 8th week body wt. in the progeny.
7. Average egg production in a flock of white leghorn is 180/year. Birds are selected from the flock and average performance of the selected flock is 240 egg/syear. Heritability of egg production is 0.3. What is expected progress in next generation?
8. Mean weaning weight of sheep is 14.5 kg. Mean weaning weight of animals selected for breeding is 16.2 kg. Calculate the SD and the expected genetic progress in the next generation. $h^2 = 0.33$.
9. In a herd of cattle, the average one year weight was 800 pounds. Males

and females were selected for breeding with the average weight of 1000 lb & 825 lb respectively. The average one year weight in the next generation was 845. Estimate the realised h^2.

10. The mean weaning weight of Nilgiri flock is 14 kg. Those selected for breeding has an average of 16.5 kg. The performance of progeny is 14.8 kg. Calculate the SD and realised heritability.

11. In a herd of 50 cows the 1st lactation yield was averaged 1500 kgs. Let us assume that 30 cows were selected as breeding stock and they average 2500 kg. What will be the response if h^2 of milk yield is 0.3.

Chapter 34

Inbreeding and Cross Breeding

The second way opened to a breeder to change the genetic constituent of the population is the mating system i.e. inbreedng and cross breeding.

Inbreeding depression:

Inbreeding is adopted for the two main purposes.

Production of lines for subsequent crossing to get hybrid vigour.

Production of uniform strains particularly of lab animal for use in research work.

However inbreeding depresses many traits and cause deterioration of vigo ur and health. This is due to increased proportion of homozygotes at the expense of heterozygotes. The proportion of heterozygous individuals decreased by 2 pqF in a population with the inbreeding coefficient of F. i.e. 'F' of 0.5 indicates that 50% of the loci that were heterozygous in the base generation have become homozygous. The reduction of the mean phenotypic value shown by the reproductive capacity or physiological efficiency due to inbreeding is known as inbreeding depression. The change of mean value resulting from inbreeding is $-2F\,pqd$ where p and q are the gene frequencies in the population, d - represents the degree of dominance and F - represents the inbreeding coefficient. This shows that a locus will contribute to a change of mean values on inbreeding only if d is not zero i.e. AA genotype differs from Aa. The direction of change is towards the value of the more recessive alleles. The equation also shows that the magnitude of change of mean depend on the gene frequencies. It is maximum when $p = q = 1/2$. Genes at intermediate frequencies therefore contribute more to a change of mean than genes at high or low frequencies when loci combine additively, the change of mean on inbreeding should be directly proportional to the inbreeding coefficient.

Effects of inbreeding:

There are differences among the traits in sensitivity to inbreeding. Traits like butter fat percentage in cattle, back fat thickness in pigs, and egg size in poultry shows little or slightly positive change under inbreeding. The most sensitive characters are those directly concerned with reproductive rate or fitness. Litter size in swine, egg number and hatchability in poultry and milk yield in dairy cattle are more sensitive characters. Growth rate is highly sensitive to inbreeding.

Depression of performance and other damages due to inbreeding are not the only results of homozygosis created by inbreeding. Different alleles become homozygous in different lines coming from the same base population. Therefore as inbreeding continues, genetic differences become larger between lines or families and the genetic variance within lines or families decreases.

Heterosis:

Heterosis is the phenomenon in which progenies of crosses between inbred lines are better than the average of the two parental populations, or exceed even the better of the two parental populations. Thus heterosis is the complement of inbreeding depression and usually appears in traits that show depression of performance under inbreeding. The cause of heterosis is the dominance effect. It is absent when the traits are influenced only by additive effects. Characters subject to inbreeding depression and showing dominance in crosses are called as heterotic characters.

Consider a population subdivided into a number of lines. If the lines are crossed at random, the average inbreeding coefficient in the crossbred progeny reverts to that of the base populations. Thus if the number of crosses are made at random between the lines the mean value of any character in the crossbred progeny is expected to be the same as the population mean of the base population, i.e. heterosis on crossing is expected to be equal to the depression on inbreeding in the absence of selection. Further if random mating is followed among the crossbreds, the inbreeding coefficient as well as population mean will remain unchanged.

Example: 30 lines taken from base population of mice were inbred by 3 consecutive generation of full sib mating.

The litter size before inbreeding = 8.1
inbred = 5.7
crossbred = 8.5

The amount of heterosis expected in crossing is dy^2, where d refers dominance and y refers to the difference in gene frequency between two lines or populations. The heterosis depends upon dominance for its occurrence. Loci without dominance, where $d = 0$ cause neither inbreeding depression nor heterosis. Heterosis will be greater when one allele is fixed in one population and the other allele in other populations. Heterosis in other words is the increased fitness of heterozygous individuals.

The dominance theory of heterosis:

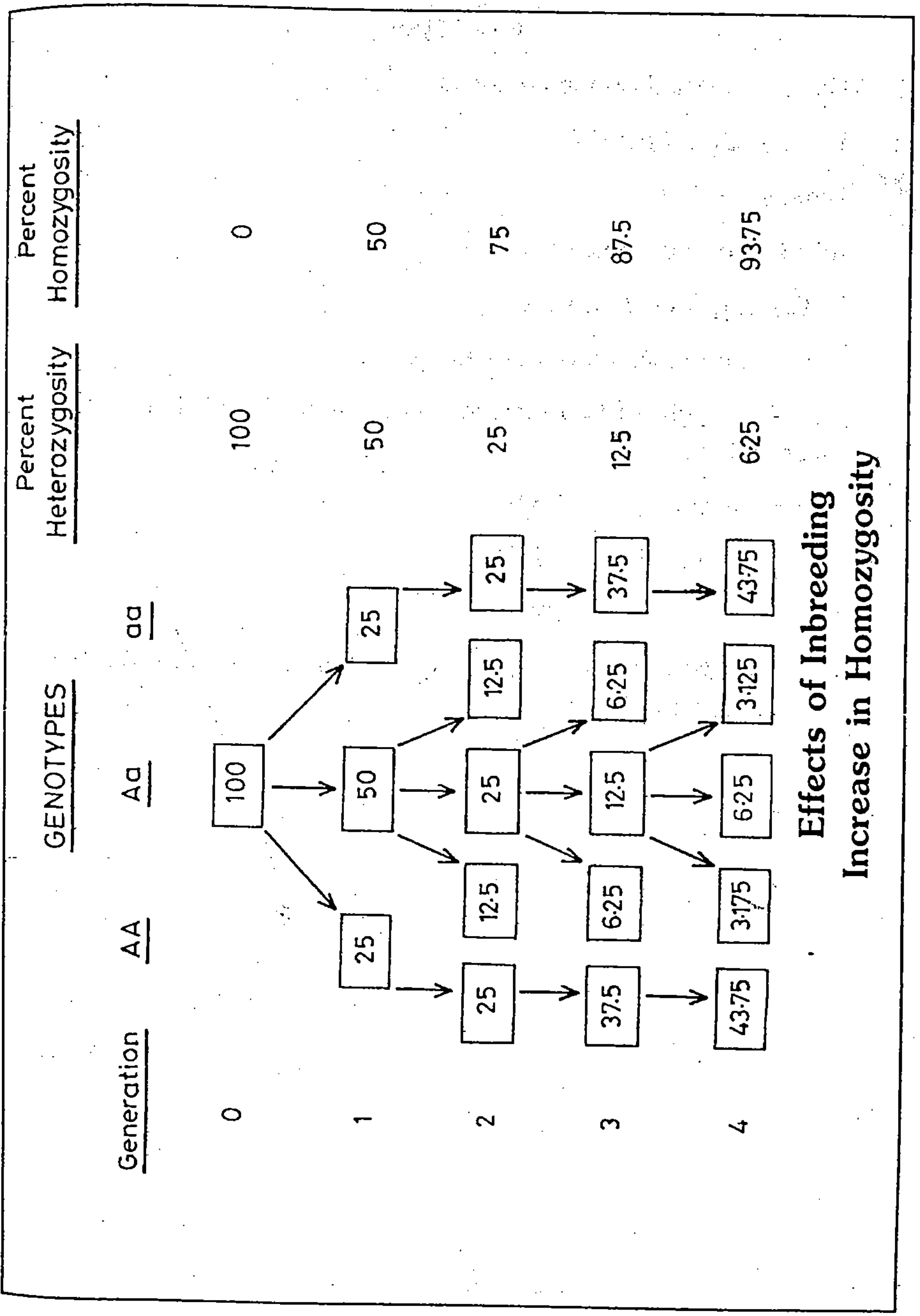
Heterosis is due to action and interaction of dominant factors.

Eg: Assuming four loci are contributing a quantitative character, each recessive genotype contributes one unit and each dominant genotype contributes two units to the phenotype. Crosses between inbred lines could produce highly heterotic F_1 than other parental lines.

AA bb CC dd X aa BB cc DD
2 + 1 + 2 + 1 | 1 + 2 + 1 + 2
= 6 | = 6
Aa Bb Cc Dd
2 + 2 + 2 + 2
= 8

The heterozygotes are less subjective to parental influences than homozygotes. Heterosis is also due to overdominance.

Cross made at random between inbred lines without selection are expected to have a mean value equal to that of the base population. Thus the inbreeding and cross breeding alone can not be expected, to lead to an improvement, but must be supplemented by selection. In practice, some improvements can be expected due to natural selection. Natural selection eliminates lethal and deleterious genes which may lead to some improvement. But this will not be great because the deleterious and lethal genes will be in lower frequency in the population. Therefore the bulk of the improvement must come from the artificial selection applied to the economic traits.


Generation
GENOTYPES
AA
Aa
aa
Percent Heterozygosity
Percent Homozygosity
0
100
100
0
1
25
50
25
50
50
2
25
12·5
25
12·5
25
25
75
3
37·5
6·25
12·5
6·25
37·5
12·5
87·5
4
43·75
3·175
6·25
3·125
43·75
6·25
93·75

Effects of Inbreeding
Increase in Homozygosity

EXERCISE

1. Discuss in detail on inbreeding depression.
2. Write essay on heterosis.
3. Write short notes on
    a) the effects of inbreeding
    b) the purposes of inbreeding
    c) the dominance theory of heterosis
4. Average height of two inbreed maize varieties and their progeny is given below:

Inbred parent -I = 35.8"
Inbred parent -II = 21.5"
F_1 = 32.4

Calculate the amount of heterosis exhibited by F_1 and what will be the average height of F_2 plants.

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Biochemical Genetics

Chapter 35

Biochemical Genetics

Gene is a minute chemical structure not visible even under the microscope. It is responsible for the occurrence of different characters almost in a specific manner. So we have been relating the genes to the occurrence of various traits. How is it possible that the gene is able to accomplish this development of traits in an orderly manner? This has been the subject of investigation by biochemists for a long time and there is overwhelming evidence to show that most of the genes exert their influence by means of enzymes, whose specificity they control.

Biochemical genetics deal with the relationship between genes and the specific biochemical reactions which lead to specific phenotypes. All biochemical reactions are catalysed by enzymes which are polypeptide chains or the proteins. The gene controls the sequence of amino acids in a protein. Number of biochemical reactions that take place in the cell resulted in the production of the phenotype. These biochemical actions are under genetic control and each reaction or process is resoluable into a series of stepwise reactions, each step is being controlled by a single gene. Alteration or mutation of a single gene results in change in the biochemical reactions.

Garrod's discovery:

A direct relationship between genes and enzymes was postulated as early as 1909 by Garrod. He described various physiological abnormalities of man in his book *"Inborn errors of metabolism"*. He observed that these abnormalities appear because of the absence of certain enzymes which are present in normal persons. He proposed that the genes produce enzymes whereas the mutant genes do not produce them, hence resulting in physiological abnormalities. He studied the metabolism of the amino acid phenylalanine and showed that the metabolism proceeds in chains of enzyme mediated reactions and a change or absence of an enzyme results in an abnormality. These abnormalities are inherited, hence the presence or absence of enzymes is under genetic control.

Phenylketonurea:

Phenylalanine is an amino acid and is part of proteins, it is released free by the breakdown of proteins in the digestive tract. Once free in the digestive tract, phenylalanine is diffused into body cells where it follows one of the following three courses:

1. It forms a part of the cell protein.
2. It is converted into tyrosine, another amino acid.
3. It is converted into phenylpyruvic acid.

The breakdown of phenylalanine is mediated by several enzymes, in the absence of anyone of the enzymes, there is incomplete breakdown which results in various metabolic disorders. Several such metabolic disorders are hereditary showing that the genes cause these disorders by failing to produce the specific enzymes.

In normal individuals all enzymes are present for the normal metabolism of phenylalanine. In one genetic condition when the recessive gene appears in a homozygous condition, there is failure to produce the enzyme which converts phenylalanine to tyrosine. In such cases there is excess of phenylalanine and phynylypyruvic acid in the blood and urine. This condition is known as phynylketonurea (PKU) and can be detected by examination of urine and plasma.

In normal persons the phynylalanine level in plasma is 1-2 mg/100 ml. In a P.K.U. patient this ranges from 16-63 mg. per 100 ml. The normal level of this substance in urine is 30-100 mg. per 100 ml. In a P.K.U. patient it ranges from 300 to 1000 mg.

P.K.U. patients have a great mental retardation. Phenylalanine interferes with the development of the brain and damage is done during the first five years of life.

64% of the P.K.U. cases have an I.Q. less than 20, idiot level. The incidence of this condition in USA is 1 out of every 25,000 children born.

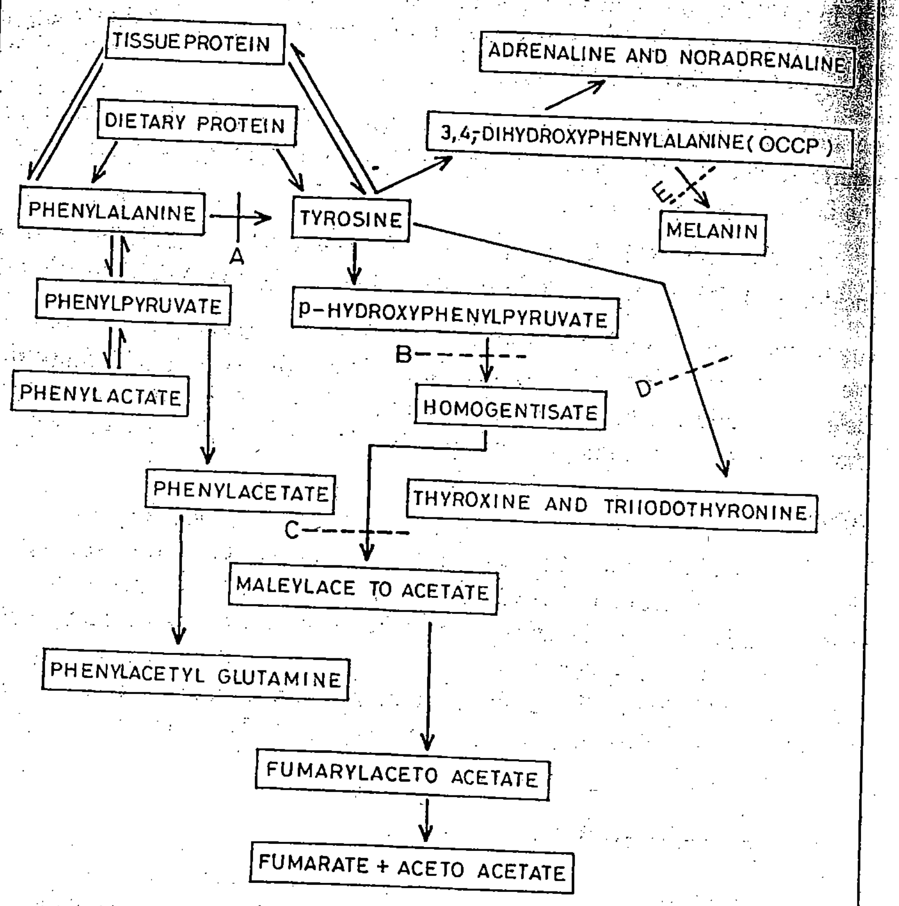
If the diet of children known to have P.K.U. during the first 5 years of life is suitably modified to contain limited amount of phenylalanine the deleterious effects can be avoided. By the 5th year full development of the brain occurs and a damage is not possible by phenylalanine later.

Albinism:

Tyrosine is another substance in the enzyme series. It may be formed directly from the protein of food or from phenylalanine. An enzyme converts it into dihydroxy phenylalanine which in turn converted into melanine, the pigment found in the skin and hair. The presence of the recessive genes 'aa' blocks the production of the enzyme needed to convert Tyrosine into dihydroxy phenylalanine. No melanin is produced. This condition is known as albinism and it is hereditary.

Genetic goitrous cretinism:

Tyrosine combines with iodine in the thyroid gland to produce thyroxine and tridothyronine. This hormone is necessary for normal physical and mental development. A pair of recessive genes 'cc' results in failure to produce the normal enzyme required to produce thyroxine and thus results in genetic goitrous cretinism. When the child shows hypertrophy of thyroid gland and the condition of cretinism, if thyroid hormone is administered and continued throughout its life, normal physical and mental development will result.


TISSUE PROTEIN
ADRENALINE AND NORADRENALINE
DIETARY PROTEIN
3,4-DIHYDROXYPHENYLALANINE (OCCP)
PHENYLALANINE
TYROSINE
MELANIN
A
E
PHENYLPYRUVATE
p-HYDROXYPHENYLPYRUVATE
B
PHENYLACTATE
HOMOGENTISATE
D
PHENYLACETATE
THYROXINE AND TRIIODOTHYRONINE
C
MALEYLACE TO ACETATE
PHENYLACETYL GLUTAMINE
FUMARYLACETO ACETATE
FUMARATE + ACETO ACETATE

Genetic Blocks in the Normal Metabolism of the Amino Acids Phenyl Alamine and Tyrosine in Human
These blocks lead to the genetic syndromes.
A - Phenyl Ketonuria B - Tyrosinosis
C -Alkaptonuria D - Goitrous Cretinism
E - Albinism

Degenerative arthritis:

Much of the tyrosin in the cell is finally reduced to CO$_2$ and H$_2$O and nitrogenous wastes with release of energy in the cells. The degradation takes place by series of enzyme action. It is converted into homogenetic acid which in turn breaks down to CO$_2$ and water. The presence of a gene 'h' in a homozygous conditions affects this conversion of hemogenetisic acid into water. The excess of this substance also called 'alkapton' (Gr. black) is excreted in urine and the person is said to have alkaptonuria. A mother can easily detect if her child has it soonafter birth by examining the baby's soiled diapers. The homegenetsic acid on exposure to air turns amer and then a black colour.

A child with this condition may develop later in life degenerative arthritis due to crystallisation of the acid in the cartilages of the body. There is darkening of the cartilages of the body most noticeable in the ears and the nose.

By following the pathway of one simple amino acid we have seen how complicated the enzyme series are and how genetic abnormalities can develop when one link in the chain of reaction is broken. Such pathways and abnormalities in respect of metabolism of other amino acids do exist.

One gene one enzyme theory:

Beadle and Tatum in 1941, developed a new experimental approach for the study of the genetic control of metabolic reactions in common bread mould, Neurospora crassa.

Extensive studies with neurospora, a mould showed that there are several genes each controlling the production of one enzyme. Wild type moulds grow in a basal medium without amino acids, since the enzymes they produce are capable of constituting the basal medium and synthesising the requisite amino acids by themselves.

When mutant forms, produced by X-rays, are grown in the basal medium they fail to grow since they do not produce one or more of the enzyme required to synthesise the amino acid concerned.

For instance when a mutant form fails to grow, but does grow when arginine is added in the basal food medium it is an indication that it has lost the power of producing the enzyme required to synthesize arginine. Similarly several genes producing specific enzymes have been recognized.

On the basis of experiments conducted on Neurospora, Beadle and Tatum proclaimed the one gene one enzyme theory. According to this, each gene has only one main function, directing the formation of only one enzyme and thus controls the single chemical reaction catalysed by that enzyme.

Fisher (1957) studied the synthesis of methionine in Neurospora and observed that the amino acid is synthesised as a chain of chemical reactions, each under the control of a single enzyme.

$$\xrightarrow{\text{Gene A}} \text{Cysteine} \xrightarrow{\text{Gene B}} \text{Cystathionine}$$

$$\xrightarrow{\text{Gene C}} \text{Homocystein} \xrightarrow{\text{Gene D}} \text{Methionine}$$

Mutations at any one of the four loci result into strains auxotroph to methionine. One mutant grows if supplied with either cystathionine, homocystein or methionine but, not if supplifed with only cysteine. The second mutant grows if supplied with either homocysteine or methionine but not if supplied with only cysteine or cystathionine or both. The third mutant grows only if methionine is supplied. Horowitz (1947) isolated number of mutants of Neurospora crassa lacking the ability to synthesise arginine.

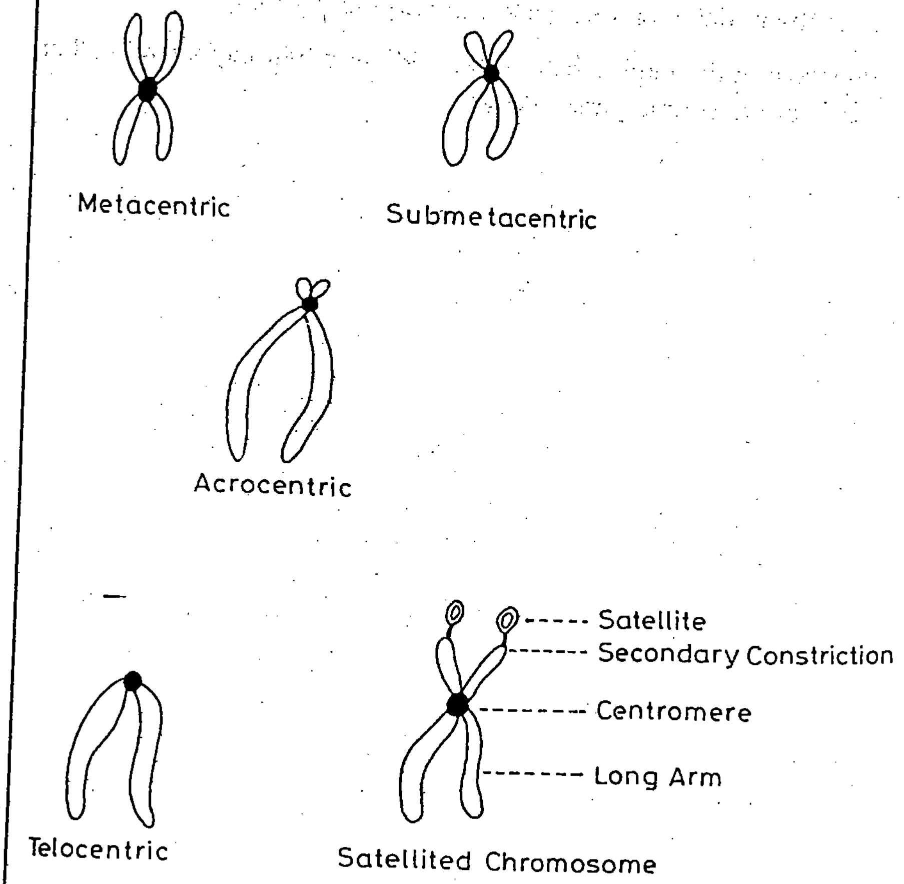
The loss of enzymatic activity caused by a mutation is referred to as genetic block. The block may be complete with the total absence of the enzyme. It may be partial in which instance enzymes are produced in insufficient quantity or produced in an altered form.

One gene one enzyme relationship in the biochemical pathway concludes that the ultimate produce of the gene is an enzyme. The gene is a segment of DNA which ultimately controls the synthesis of a protein or an enzyme. A change in the base sequence of DNA constituting a gene is reflected as a change in the amino acid sequence of the protein which in turn is reflected as change in the phenotype.

EXERCISE

1. Define Biochemical Genetics.
2. Describe on the phenylketonurea condition.
3. Describe in detail on one gene one enzyme theory.
4. Phenlketonurics tend to have lighter skin and hair pigmentation than their normal counterparts. Why?

Types of Chromosomes


Metacentric
Submetacentric
Acrocentric
Satellite
Secondary Constriction
Centromere
Long Arm
Telocentric
Satellited Chromosome

Aneuploidy in the Human Populations

Chromosome Nomenclature	Chromosome Formula	Clinical Syndrome	Estimated Frequency	Phenotypic Characteristics
47, +21	2n + 1	Down	$\frac{1}{700}$	Short broad hands with simian-type palmar crease, short stature, hyperflexibility of joints, mental retardation, broad head with round face, open mouth with large tongue.
47, +13	2n + 1	Trisomy-13	$\frac{1}{20,000}$	Mental deficiency, deafness, cleft lip polyductyly and cardiac anomalies.
47, +18	2n + 1	Trisomy-18	$\frac{1}{8,000}$	Multiple congenital malformation of organs, malformed ears, small mouth and nose, mental deficiency, double kidney short sterrum, 90% die in the first 6 months
45, X	2n - 1	Turner	$\frac{1}{3,000}$	Female with retarded sexual development, usually sterile, short stature, webbing of skin in neck region, hearing impairment, cardiovascular abnormalities.
47, XXY	2n + 1	Klinefelter	$\frac{1}{500}$	Male, subfertile with small testes, developed breasts, femine pitched voice, long limbs, knock knees, rambling talkativeness.

Chapter 36

Genetics of Microorganisms

The genetic study of microorganisms such as viruses and bacteria have many advantages. The microorganisms generally have only one set of chromosomes or one set of genome as is characteristic of haploid gamete of a diploid organism. Since only one allele is present in an individual, the genes of a haploid organism are neither dominant nor recessive. In this respect, the study of micro-organisms is relatively simple because every gene which the individual organism possesses can be expressed in its phenotype. —Further the study of rare events like mutations is easier since large populations of microorganisms can be raised in a petridish or test tube. A population upto 10^6 to 10^9 organisms can be grown in a petridish. Use of selective media can easily isolate one mutant individual cell in a million. The relative simplicity of micro-organisms such as viruses offers the best opportunity for studying gene structure and function at molecular level. Many organisms are having short generation interval. Some bacteria divide about once in 30 minutes. So within fifteen hours 30 generations can elapse, producing approximately billion progeny cells. All of the asexual descendants

Sex-chromatin: Sex difference in the staining of many somatic nuclei are also noticed. Barr and Bertram, 1940, first noticed this. Many female cell nuclei contained a dark staining spot, which was lacking in males. These bodies are known as Barr bodies or chromatin positive bodies. Their unexpected appearance or absence in many cases of sex abnormalities was noticed.

- Number of Barr bodies is usually one less than the number or of X chromosomes. Lyon and Russel proposed (Lyons hypothesis) that this represents the inactivation or heterochromatization, of all but one of X chromosomes. That is, a female with two X chromosomes will have one of these chromosomes somatically inactive, so that dosage relationship between her sex chromosomes and autosomes will be the same as in the somatic tissues of male. Individuals with more than two X chromosomes are therefore viable, since all but one of the X-chromosomes are inactivated in many of the tissues. This inactivation of additional X-chromosome is random.

Contrary to Drosophila, the Y chromosome in humans are necessary to produce the male phenotype, Xo individuals are female (Turner syndrome) although sexually non-functioning and XXY individuals are males because of presence of Y chromosome, although sterile. No matter how many additional X chromosomes are present (as many as 4 have been observed), the presence of single Y chromosome is sufficient to shift development toward mascularity. About 1.7 per 1000 children arise from gametes in which some form of sex-chromosome non-disjunction has occurred.

XO — Turner syndrome:

One out of every 3,000 female births results in a child with this abnormality. These are phenotypic females, but at adolescence the adult characteristics do not develop normally and they never reach functional maturity. Persons affected may be dwarfed physically, and show mental retardation also. Many of them carry a characteristics `webbing' of the skin on the side of neck, wide spaced nipples of the mammary glands. Ovaries in individuals carrying this defect composed mainly of masses of connective

tissue. Estrogen injection in such females may bring marked improvement in the female phenotype.

These individuals carry only one X chromosome. Non-disjunction during meiosis can produce gametes with odd sex chromosome complements and can result in these sexual abnormalities.

In case of XXX-individuals occurring nearly at one in 830 female births, are normal in appearance, may be fully fertile although most are mentally retarded.

XXY-Klinefelter syndrome:

About one male child out of every 5,000 born may express this syndrome. Such children have typical male sex organs, but as they grow, the testes do not grow proportionately to the rest of the body and at the adult stage are only about one-half of normal size. Also, the fat deposits and growth of face and hair are some what feminine in nature and some degree of breast enlargement (gynacomastia) may occur. Man with Klinefelter syndrome are always sterile and about 25% have some degree of mental retardation. The individuals carry two X chromosomes and one Y chromosome in their somatic cells.

Chromosomal abnormality related to autosome : Down's syndrome-21 Trisomy - Mongolism

Affected individuals are characterized by physical abnormalities of the face, eyelids, tongue, and other parts of the body and are greatly retarded both physically and mentally.

It is probably the most common congenital abnormality, with a frequency of one out of 600 births. It is characterized by mental retardation, a short body with stubby fingers, swollen tongue, monkey like skin ridges on the extremities, and eyelid folds resembling those of Mongolian races. Higher incidence is reported among children born of older mothers but not of older fathers.

EXERCISE

1. Describe in detail on genetic recombination in bacteria.
2. Write briefly on the following:
    a) Advantages of bacterial genetics
    b) Transformation
    c) Conjugation
    d) Transduction
    e) Sexduction

Immunogenetics

Chapter 37

Immunogenetics

Immunogenetics is the science which has union of the two disciplines immunology and genetics. It consists of the use of immunological tools for the detection of genetically controlled variation. It is the general area of knowledge associated with the genetic basis of immunity.

Immunogenetics was born during the early periods of this century when Landsteiner discovered the ABO blood groups in human beings. In the earlier years immunogenetics originally consisted of the determination of blood group or blood typing. Then it was extended to include all the genetic variation detected by immunological means.

Blood group antigens:

Blood typing is a characterisation of structures on blood cells and a determination of differences among soluble macromolecules of the blood. It may be just like fingerprints. Since the structures being characterized or differentiated are under genetic control, they are often referred to as genetic markers. They do not change throughout one's life. Hence the classification of blood cells is permanent. Genetic markers on red blood cells are called blood group factors.

EXERCISE

1. Define Immunogenetics, and its application.
2. Write in detail on blood group antigens.
3. Write briefly on Phenogroups.
4. Write short answer on neonatal isoerythrolysis.
5. Discuss on major histocompatability complex.

Developmental Genetics

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पुस्तक खराब करु नये.

Chapter 38

Developmental Genetics

Two words are used essentialy in Biological science. Theory and Explanation. Theory is more aptly termed as hypothesis. Explanation is an amorphous concept and means any statement.

In case of development we can look up at it and try to explain in molecular terms and looked down to explain in evolutionary terms. Molecules are at the smallest level. Development is in the midway and evolution is at higher level. During last two decades, Developmental science has become totally transformed due to the staggering success of Molecular biology and Molecular genetics.

Molecular geneticist believes that all the substances that make up living organs are derived from DNA. DNA gives rise to specific RNA which in turn designates specific proteins, many of them are enzymes that control specific organic reactions that are taking place in the cells. As per this accepted view protein can not make DNA or other protein for that matter. All the basic instructions for the molecular composition of organism comes from DNA. Developmental biologists doubt in terms of what are the instructions that make an egg into a plant or animal and they are satisfied that they do not all sproud immediately from

DNA. There are many other things such as the polarity of egg, the distribution of special chemicals in cytoplasm, the structure of egg cortex none of which seems to be have direct connection with the DNA. But according to Molecular biology, there is a link between substances in cell and DNA.

Three kinds of events occur in development.

1. Synthesis of substances.
2. Timing of their synthesis.
3. Localisation of substances.

The role of DNA in the synthesis is more obvious but is less so in the process of timing and the localization. But ultimately DNA is responsible for it.

Not all products of DNA are made new in each life cycle. Many products are transmitted from parents life cycle. The statement that DNA is responsible for the synthesis of specific substances and their localization can be true only if one adds up numerous life cycles.

There are two kinds of development which are appearing simultaneously.

1. Evolutionary development.
2. Life cycle development.

The life cycle development are in general connected one to another by a single celled stage namely a fertilised egg or an asexual spore.

Evolutionary development are connected to natural selection. Selection can not operate without reproduction and inherited variation without making of copies.

Life cycle can never be diversed from natural selection. In the beginning they may simply have involved nucleic acid replication but as protein came to be produced by and associated with nucleic acid, eventually an entire prokaryotic cell was constructed and this became the minimum unit or link between one life cycle to that of the next.

The growth and development of an individual from an egg involves a gradual differentiation, an increase of diversity. Such development is termed as Epigenesis. The ways in which genes may act in affecting characters are many and varied. Genes may affect the cytoplasm of the egg in the initial stages or final stages of development. They may modify the direction or the plane of cell division. They may produce an enzyme at a particular stage of development or they may fail to produce it.

In order to devleop from egg to adult growth is necesssary. There is synthesis of new protoplasm and it manifests itself in the form of cell division and cell enlargement usually occurring simultaneously. In organisms that increase in size by growth, there is often a shapping of the embryo which is generally termed as Morphogenesis.

In these growing and shaping of masses of protoplasm there is ultimately a difference in different parts, a difference in organells, cell or tissue structure which is known as cell differentiation. During Morphogenesis, the phenotype changes radically and this is known as phenogenesis. Phenogenesis concerns not only early life but also extends to adult form and senescence.

The developmental process is initiated by a major gene and regulated through the control of major genes action. The end of the process is marked by the production of phenotype. A phenotype may be anything along the course of developmental process. The phenotype could be first gene product in which case developmental process would be a short one, i.e. the connection between process initiated by gene and phenotype is a very direct one. The DNA structure of gene itself could be taken as a phenotype.

We may summarize the present knowledge by stating that a complete complement of chromosomes as well as normal environment is necessary for development.

EXERCISE

1. What are the events occurring in development?
2. Discuss on the kinds of development.
3. Write briefly on
    a) Life cycle development.
    b) Evolutionary development.

Applied Genetics

Chapter 39

Genetic Counselling

The principles of Genetics can be applied not only to obtain new or better strains of animals or plants but can be extended to human beings also. An overall progressive advance in biological science, particularly in molecular genetics has opened new exciting possibilities for the welfare of mankind. Much more foreseeable is the application of genetics to solve some of the more pressing problems of detection and treatment of genetic diseases.

Genetic counselling:

The term genetic counselling is applied to service rendered by which the prospective parents are provided with the estimates of the probability that they will produce children with the genetically controlled defects. A voluntary restriction of child birth with the inherited disorders can be brought about through proper counselling by geneticists.

Genetic counselling provides and interprets medical information based on our expanding knowledge of human genetics. One area of specialization is biochemical genetics which makes use of special tools such as electrophoresis for enzyme assay. Another area cytogenetics has defined a wide range of chromosome irregularities that have practical significance for diagnosis of human disease. Somatic cell hybridization studies brought a rebirth in linkage studies which will pave way for gene mapping. Popualtion genetics with its relations involving changes in allele frequencies and interactions with environments provides a broad theoretical base for clinical medicine.

The most common condition under which people obtain a genetic counsellor's advice is the one in which phenotypically normal couple produces a first child suffering from a major defect. They wish to know what are the chances for the subsequent children to be affected. There are quite a number of tasks for the genetic counsellor to undertake.

The first task is studying the pedigree of the couples concerned. He has to estimate the probability of genetic defects among their progeny by applying the principles of inheritance to the information already collected. He can not predict anything with certainty but only the chances of occurrence of defects. Invariably the final decision is left to the couple themselves.

Genetic counsellor must also be able to give advice to populations which have a high incidence of certain specific diseases. For example African people have higher incidence of sickle-cell anaemia and those of Mediterranean descent are prone to risk of thalassaemia. Hence Genetic counsellor must have thorough knowledge of human genetics. He should be able to advice people with an understanding of the genetic problems they have in their families. He should try to wash away the guilt feelings of the parents of the affected child by pointing out that genetic calamities can occur in any family and no one can escape all the possible undesirable expressions of his genotype.

Simple inheritance:

The genetic counsellor's task is easier in cases in which the mode of inheritance is simple. A case may come to the counsellor where the normally pigmented parents having an albino child wish to know the probable chances of their next child also being albino. The child is homozygous for albino gene, and hence both the parents must have contributed one allele each to the child. In such cases, no further pedigree analysis is required and the probability of the child being albino is one out of four or 25% on the basis of the principles of simple recessive inheritance. It is for the parents to decide

whether they would undertake this risk of 25% or not.

Complex inheritance:

The genetic counselor's task becomes more complicated. When the pedigrees fail to establish simple mode of inheritance. For example congenital dislocation of hip in twins which is influenced by non-genetic factors.

The task of genetic counselor is not easy. In many occasions, he has to make decisions on vague only. One of the most difficult tasks is the preparation of accurate pedigree reports. The problem is more complicated with complex psychological characteristics like neurotic behaviour. The role of genetic counsellor will become more important with the perfection of the techniques of genetic engineering.

EXERCISE

1. Define genetic counselling and discuss on its application.
2. List out the possible reasons for genetic disorder in human beings.
3. What are the possible approaches for genetic quality control?
4. Why is the knowledge of family record so essential in the genetic counselling?
5. What are the functions of a genetic counsellor?
6. Discuss on the usual difficulties that are likely to be encountered by a genetic counsellor.
7. What are the limitations of genetic counselling.

Chapter 40

Genetic Engineering

Genetic engineering is defined as the science dealing with the correction of nature's mistakes at their source the DNA molecule. Through its techniques it carries out the modification of specific genes or implants desired new genes into chromosomes.

Genetic engineering can also be defined as the technique of adding removing or replacing genetic units in order to achieve permanent and heritable changes in plants or animals for the welfare of the mankind. Terms such as 'gene surgery' gene therapy, gene manipulation and gene transplantation have been used synonymously with genetic engineering.

Certain genetic disorders are due to a wrong nucleotide sequence which codes for a wrong amino acid chain. Even a replacement of one base pair or insertion of one extra base pair, or the deletion of one base pair from a gene will result in altered function of the gene. How such defective nucleotide sequences could be replaced by the normal ones is the problem in genetic engineering.

Gene isolation and transformation are the procedures involved in genetic engineering. DNA fragments are obtained by bursting the cell wall, separating other substances of cell and by cutting the DNA. Then the DNAfragment is inserted into bacterial DNA. Mixing of DNA with bacterial DNA can be done in two ways. It may be mixed with bacterial DNA or alternatively they can be spliced into the DNA of a virus which then in turn inserts itself into the bacterium DNA. A whole colony of bacteria for each of the DNA fragments thus develops after sometime. Then each colony of bacteria is tested to identify the desired gene fragment.

Transformation:

Griffith observed that extracts of the heat-killed virulent pneumonia bacteria injected into mice together with living non-virulent bacteria turned the latter into virulent bacteria as if a real hybridisation had occured.

Griffith's experiments which led to the discovery of DNA as the genetic material represent one of the major techniques of genetic engineering. In this technique the isolated DNA is incorporated into the genome of a cell which is incubated along with the donor cell.

The phenomenon of transformation is of fundamental significance to genetics since it occurs solely at a molecular level.

Synthesis of gene:

One of major achievements of the recent years in molecular genetics is the artificial synthesis of gene coding for yeast alanine transfer RNA and bacterial tyrosine transfer RNA by Noble lanrete H.G. Khorana during the year 1968.

In Khorana's experiments DNA fragments of 10 to 20 nucleotides in length were synthesised and these fragments were joined enzymatically increasing the chain size by 5 to 6 units at a time. He synthesised structural gene containing 78 nucleotide units in each chain and also synthesised chemically small nucleotide bits of chain length varying from 6 units to 20 units aligned them in correct order and joined the bits enzymatically using ligase enzyme. The gene has unique start and termination points. The start signal is present on the left side and termination signal is present on the right side of the gene to be transcribed. The gene was made to work by introducing it into the DNA of a virus that lines in E.coli. Before the introduction of gene, the virus was unable to grow in bacteria. After introduction the virus was able to grow. It expressed as if it were a part of the bacterial Genotype.

Recombinant DNA Technology:

Stanley Cohen and Herbert Boyer (1973) discovered the Recombinant DNA technology or gene splicing which makes it possible to cut out precisely defined segments of DNA from one organism and splice them into the genosome of another. The experiments for creating recombinant DNA are engineering experiments, since cutting and slicing of DNA molecules are being carried out very much in the manner one cuts and joins tapes.

The recombinant technique involves the plasmids and two groups of enzymes called endonucleases and ligases. Plasmids the extra segments of DNA are present in the cytoplasm of cells. The genes of plasmids code for certain substances which impart antibiotic resistance to cell. Plasmids are of very small size and can be isolated from one cell and transferred to another cell. A group of enzymes known as restriction endonucleases can cause a cut a specific site in the DNA segment. The cut results in sticky ends at either side of the DNA molecule. Any desired segment can be introduced into the cut, since sticky ends of the DNA molecule can join to any other segment of the DNA. Introduction of the desired segment of DNA is done with the help of another set of enzymes - ligases. Ligase function as a sealing enzyme. This modified plasmid is then either introduced into the original bacteria or ~~in~~ any other strain of bacterim. The newly joined material ~~is~~ known as recombinant DNA. The host bacterium will follow the instructions of foreign DNA just as if it were its own. The bacterium multiplies and continue to multiply normally reproducing foreign gene among their own genes. The repeated division of the bacteria with new gene results in a colony of identical cells known as clones. This process of getting many identical copies of genetic material from one bacterium through multiplication of bacteria is known as cloning.

Recombinant DNA technology is applied to map the chromosomes in fundamental genetical research for detection of diseases in medical field and for higher yield in agriculture. Genetic engineering not only has potential benefits but also has contain destructive effects. There is a positive danger that the manipulation of genes might be accident result in the origin of new kinds of diseases and organisms containing fatal genetic elements. By replacing the defective nucleotide sequences with the normal ones it is possible to cure many hereditary diseases. Changing the genome by genetic engineering will give plants and animals of desired gene pool and human beings with predetermined characteristics are possible.

EXERCISE

1. Define genetic engineering.
2. Discuss on the practical applications of genetic engineering.
3. Write short answers on
   a) Transformation
   b) Synthesis of gene
4. Write in detail on Recombinant DNA Technology.
5. What are the important techniques employed in Genetic Engineering.
6. Discuss on cloning and its uses.

Glossary

Glossary

Aberration	Variation in the structure and/or number of chromosome.
Acentric	A fragment of chromosome without a centromere.
Acquired characters	The alteration in the morphology and or functions of an organism in response to environment. Acquired characters are not heritable.
Acrocentric	A chromosome or chromatid with a nearly terminal centromere. One arm of chromosome is very short.
Adaptive value	Fitness of the individual.
Additive genes	Non-allelic genes affecting the same trait with cumulative effect on the phenotype.
Adenine	A purine base found in RNA and DNA. Pairs normally with thymine in DNA.
Agglutinin	An antibody in blood plasma that brings about clumping (agglutination) of the blood cells that carry an incompatible agglutinogen.

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Agglutinogen	An antigen in red blood cells that reacts with a specific agglutinin in the plasma and causes clumping of the cells.
Albinism	Absence of pigment in skin, hair and eyes of an animal. Absence of chlorophyll in plants.
Alcaptonuria	An inherited metabolic disorder. The affected individual excrete excessive amounts of homogentisic acid (alcapton) in the urine which turns black when exposed to air.
Allele	A gene which can occupy the same locus as another gene in a pair of homologous chromosomes.
Allopolyploid	A polyploid having chromosome sets from different species.
Allotetraploid	An organism with four sets of chromosomes derived from hybridization of different species.
Amino acid	The basic structural units of proteins. These are a class of organic compounds containing an amino group (-NH_2) and a carboxyl (-CooH) group, plus a side chain.
Amphidiploid	A species of plant derived from doubling the chromosomes in the F_1 hybrid of two species; an allopolyploid. In an amphidiploid the two species are definitely known. Whereas in other allopolyploids they may not be known.
Anaphase	The stage of nuclear division in which the daughter chromosomes move from spindle equator to spindle poles of the cell. This follows metaphase and precedes telophase.
Anaemia	A deficiency of haemoglobin or reduced number of erythrocytes resulting in abnormal condition characterised by pallor, weakness and breathlessness.
Aneuploidy	Variation in chromosome number but less than an entire set 2n+1 (Trisomy); 2n-1(monosomy).
Anomalous gametes	Gametes having chromosomes with irregular morphology or numbers.
Antibody	A substance that neutralises a specific antigen (foreign substance) in a living organism.
Anticodon	Three bases in a transfer RNA molecule that are complementary to the three bases of a specific codon in messenger RNA.
Antigen	A substance usually a protein that stimulates the production of antibodies when introduced into a living organism.
Artificial selection	The process of choosing the parents of next generation.
Asexual reproduction	The process of reproduction that does not involve the union of gametes from the two sexes.
Assortative mating	A mating in which similar types tend to pair. Non-random mating.
Asynapsis	The failure or partial failure in the pairing of homologous chromosomes during the meiotic prophase.
Atavsim	Reappearance of an ancestral but not parental trait after several generations. A gene comes to expression after a period of non-expression because of recessiveness or other masking efects. Also known as reversion.

Centrosome	-	A self-propagating cytoplasmic body usually present in animal cells that consists of a centriole and sometimes a centrosphere (when inactive) or astral rays (when active). It is located at each pole of the spindle during the process of nuclear division (mitosis).
Character	-	One of the many details of structure form, substance or function that make up an individual. It is the outcome of genotype and environment interaction.
Charged tRNA	-	tRNA carrying an amino acid.
Chiasma	-	The visible connection or crossover between two chromatids observed during prophase I of meiosis (PI-chiasmata).
Chimera	-	An individual whose cells are of two or more genetically different constitutions. It may result from different zygotes that grow together.
Chi-square test (X^2)	-	A statistical test used to determine if observed frequencies differ from those expected on the basis of a specific hypothesis.
Chloroplast	-	Plastid containing chlorophyll, site of photosynthesis in green plants. A mode of cytoplasmic inheritance independent of nuclear genes has been associated with these cytoplasmic structures.
Chromatid	-	One of the two daughter strands of a duplicated chromosome that are connected by a single centromere. A half chromosome results from the replication of chromosomes during a nuclear division.

Chromatin	-	The nucleoprotein of chromosomes that stain strongly with basic dyes (DNA, RNA, histones, and non-histone proteins).
Chromomere	-	A concentrated chromatin bead on eucaryotic chromosome resulting from local coiling or thickening of a continuous thread.
Chromosome	-	Nucleoprotein bodies that dark staining with basic dyes, microscopically observable in the cell during cell division. They carry the genes which are arranged in linear order. Chromosome number is constant for each species.
Chromosome arm	-	A part of a rod chromosome to one side of centromere.
Chromosome aberration	-	Abnormal structure or number of chromosomes.
Chromosome banding	-	The occurrence of light and dark areas along the length of chromosomes as a result of differential staining.
Chromosome segregation	-	Separation of the members of a pair of homologous chromosome in a manner that only one member is present in the gamete.
Cis-arrangement	-	The location of two closely linked mutant alleles in one chromosome with two normal alleles in its homologous chromosome. Also known as coupling.
Cistron	-	A unit of function in a DNA molecule, a segment of DNA specifying one polypeptide chain in protein synthesis structural gene.
Clone	-	A line of cells derived from a single cell. All cells of a clone remain genetically identical.

Cloning	Making identical copies of proteins, genes, nuclei, cells or individuals by an asexual, biological process.
Codominant genes	Alleles, each of which expresses an independent effect and contributes to phenotype when heterozygous.
Codon	A set of three nucleotides that is specific for a particular amino acid in protein synthesis.
Coefficient	A number expressing the amount of some change or effect under certain conditions (Eg. Inbreeding coefficient).
Co-enzyme	A substance necessary for the activity of an enzyme.
Coincidence	The ratio of observed double cross overs to expected double cross overs and expressed as number. It is a measure of interference.
Colchicine	An alkaloid derived from the autumn crocus that is used as an agent to arrest spindle formation and interrupt mitosis.
Combining ability	Potential to produce of high proportion of desirable hybrid individuals.
Competence	Ability of a bacterial cell to incorporate DNA and become genetically transformed.
Complementary genes	The genes which complement each other to produce a phenotypic effect.
Complementation test	Introduction of two mutant chromosomes into the same cell to determine whether the two mutations occurred in the same gene.
Compound cross	Combining desirable genes from more than two inbred lines.

Concordance	The expression of a given trait by a pair. For example identical twins both expressing the same syndrome.
Congenital defect	A defect present at birth. It may be determined genetically or by external influences.
Conjugation	Union of morphologically similar gametes or unicellular organisms during fertilization allowing passage of genetic material from donor cell to the recipient cells.
Consanguinity	Individuals related by descent from a common ancestor. Usually not further back than three or four generations.
Constitutive heterochromatin	Chromatin which is always heterochromatic. Composed of highly repetitive late replicating DNA.
Continuous trait	Quantitative trait, multigenic or polygenic inheritance represent this type of variation which can not be represented by discrete classes and it requires measurement data.
Copy choice	An explanatory term for crossing over given by J.Belling in 1930 and he suggested that crossing over occurs during the process of chromosome duplication or copying.
Coupling	Dominant alleles at two different loci are said to be in coupling in a heterozygote if they are located on the same chromosome. Thus the double heterozygote AB/ab is said to be in coupling while Ab/aB is said to be in repulsion.
Cavariance	Simultaneous variation among traits.
Cross breeding	Mating between member of different breeds.

Criss-cross inheritance	Parents of one generation passes on the sex linked character to the opposite sex in the next generation.
Crossing over	The process of exchange of genetic material between two homologous chromosomes, presumed to occur through breakage of both chromosomes at homologous sites followed by reunion after exchange.
Crossover unit	The frequency of exchange of one per cent between the two pairs of linked genes. One per cent of crossing over is equal to one unit on a linkage map.
Cytogenetics	Study of chromosomes and their implications in genetics. Study of cellular structures and mechanisms associated with genetics.
Cytokinesis	The division of cytoplasm during cell division.
Cytology	The study of the structure and function of the cell.
Cytoplasm	The protoplasm of the cell surrounding the nucleus in which cell organelles are located.
Cytoplasmic inheritance	Extrachromosomal or non-Mendelian inheritance by structures in the cytoplasm rather than the nuclear genes.
Cytosine	A pyrimidine base found in RNA and DNA. It pairs with guanine in DNA.
Darwinian fitness	The fitness of a given genotype in a given environment. It depends on both fertility and survival.
Degenerate code	The genetic code in which more than one codon may represent the same amino acid.

Deletion	The loss of a part of a chromosome involving one or more genes.
Deoxyribonucleic	Double stranded helically coiled nucleic acid acid (DNA) molecule.
Deoxyribose	The 5 carbon sugar in DNA.
DNase	Deoxyribonuclease - an enzyme that breaks down DNA.
Deuteranopia	Green blindness - sex linked recessive character.
Deviation	A variation from an expected number.
Diakinesis	The last stage in the prophase of meiosis immediately before the disappearance of nuclear membrane, in which bivalents are shortened and thickened.
Dicentric	A chromosome or chromatid having two centromeres.
Differentiation	Modification of different parts of the body for particular functions during development of the organism.
Dihybrid	An individual heterozygous in two pairs of alleles.
Dimorphism	Two different forms in a group as determined by such characteristics as sex, size or colour.
Diploid	An individual or cell having two complete set of chromosomes.
Diplotene	The stage of prophase of meiosis following the pachytene stage, but preceding diakinesis, in which pairs of chromatids derived from homologous chromosomes begin to separate

	from each other except at certain points of connection (chiasmata) where crossing over has occurred between chromatid segments.
Disassortative mating	Random mating.
Discontinuous variation	Variation in which the trait can be classified into two or more easily distinguishable classes eg. tall and dwarf plants.
Discordant	Twins are said to be discordant with respect to a trait, if one shows the trait and the other does not.
Disjunction	Separation of homologous chromosomes at anaphase of cell division.
Dispersive forces	The change in gene frequency due to sampling error.
Dizygous twins (DZ)	Twins that arise from two different eggs fertilized by two different sperms.
Dominance variance (V_D)	That portion of the genetic variance that results from the dominance effect of genes.
Dominance	An allele manifesting its phenotypic effect also in the heterozygotes, a trait determined by a dominant allele.
Dominant lethal	Action of a gene lethal in both the homozygous and heterozygous condition.
Dosage compensation	The mechanism by which genes on the X chromosomes have the same physiological effect in females who have two x chromosomes as in males who have only one x chromosome.
Double backcross	Mating of an individual who is heterozygous at two loci with an individual who is homozygous recessive at the same two loci.
Double cross over	Occurrence of crossing over at two points on the chromosome.
Double helix	Shape of the DNA molecule. Two spiral strands coiled about each other.
Duplication	Repetition of a segment in the same chromosome.
Downs's syndrome	Trisomy 21– Mongolism – A syndrome characterised by mental behavioural and physiological defects caused by presence of extra chromosome in 21st pair in human.
Electrophoresis	The migration of suspended particles in an electric field.
Embryo	The developing zygote – a young organism in the early stages of development. In humans, the first period in uterus.
Endonuclease	An enzyme that breaks strands of DNA and thus is involved in replication and recombination of DNA.
Endoplasmic reticulum	Network in the cytoplasm to which ribosomes adhere.
Endopolyploidy	Occurrence of cells in diploid organisms containing multiples of the 2n chromosomes i.e. 4n, 8n, etc.
Environment	Secondary factor influencing development of an organism and trait.
Enzyme	A protein that accelerates a specific chemical reaction in a living organism.
Episome	A genetic element that may be present in a given cell, either on a chromosome or in the

		cytoplasm. Example - For Fertility factor in E.coli.
Epistasis	-	The suppression of the action of a gene or genes of one pair by a gene of non-allelic pair. The masked gene is said to be hypostatic.
Equational division	-	Mitotic division. Also known as homotypic division.
Equational plate	-	The figure formed by the chromosomes in the centre (equational plane) of the spindle in nuclear division.
Erythrocytes	-	Red blood cells.
Erythroblastosis toetalis	-	The blood of the child contains immature nucleated RBCs causing death due to Rh factor.
Euchromatin	-	Parts of chromosomes that carry genes and have characteristic staining properties. Opposite of heterochromatin.
Eugenics	-	The application of the principles of genetics to the improvement of mankind.
Eukaryote	-	A higher organism whose cells contain a distinct membrane bound nucleus and cytoplasmic organelles which undergoes meiosis. Opposite of Prokaryote.
Euploidy	-	Variation in chromosome number by whole sets or exact multiples of haploid number e.g. diploid, triploid etc. Opposite of heteroploid and aneuploid.
Exonuclease	-	An enzyme that digests DNA beginning at a terminal nucleotide.
Expressivity	-	Degree of expression of a trait controlled by a gene.

Extrachromosomal	-	DNA units in the cytoplasm that control cytoplasmic inheritance. Structures that are not a part of the chromosomes.
F_1	-	The first filial generation, usually the progeny of a cross between two homozygous parental types.
F_2	-	The second filial generation resulting from the matings of two F_1 individuals.
F factor	-	Fertility factor in a bacterium. In the presence of F+ a bacterial cell function as a male.
Feed back inhibition	-	End product stops protein synthesis. The end product affects an initial enzymatic steps in the biosynthetic pathway.
Fertility	-	Ability to produce offspring.
Fertilization	-	The fusion of male gamete - sperm with a female gamete — ovum to form a zygote.
Fitness (W)	-	Adaptive value. The proportionate number of offsprings produced by an individual as compared with the average of population. It ranges from 0 to 1.
Fixation	-	Chance fluctuation of members of a pair of alleles around the mean that may result in homozygosity of one or the other allele (AA or aa) and therefore will prevent segregation between the different lethals.
Gamete	-	A mature male or female reproductive cell, such as sperm or egg used in fertilization.
Gametogenesis	-	The formation of the gametes.
Gametophyte	-	The phase of the plant life cycle that bears the gametes. Cells have haploid chromosomes.

Gametic selection	Differential survival of sperm or ovum.
Gene	A functional unit of genetic material consisting of nucleotides, a unit of heredity, located in a fixed place on the chromosome.
Gene action	The way in which a gene or genes control the expression of character.
Gene flow	The transfer of genes from one breeding population to another due to migration which may result in changes in gene frequency.
Gene frequency	The proportion of given allele in the population.
Gene isolation	Pulling of a single gene out of millions in a chromosome by using natural scissors - endonucleases.
Generalised transduction	Unrestricted transduction, transduction in which transducers do not have a specific site of attachment to the chromosome and have equal probability of transducing any gene.
Gene pool	The total of all genes in a population at a given time.
Gene splicing	Joining of two gene segments cleaved through the action of restriction endonucleases, by interweaving of strands at two ends with the help of ligases.
Genetic code	The code that relates nucleotide sequences in nucleic acids to amino acid sequences. Transcription of nucleic acid information in the form of mRNA codons into corresponding information of amino acid sequence in protein synthesis.

Genetic drift	In small populations, the tendency for heterozygous gene pairs to become homozygous for one allele or other by chance rather than by selection.
Genetic engineering	Directed and artificial alteration of genetic makeup of cells either through modification of specific genes or by implantation of new genes into chromosomes.
Genetic equilibrium	A condition in which a particular gene frequency remains constant from generation to generation.
Genetic load	The mean number of lethals per individual in a population. The proportional reduction in the fitness of optimal genotype of an individual or population due to the presence of deleterious genes.
Genetic marker	A gene mutation that has phenotypic effects useful for tracing the chromosome on which it is located.
Genetic polymorphism	Two or more discontinuous variants frequently occurring in a population eg. human blood groups.
Genetics	The science of heredity and variation.
Genetic screening	A systematic testing of individuals to ascertain potential genetic abnormalities in them or in their progeny that may require prophylaxis or treatment.
Genome	A complete haploid set of chromosomes and hence of genes inherited from as an unit from one parent.

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Genotype	-	The genetic make up or constitution of an individual.
Germ cell	-	A reproductive cell - gametes or their precursors. Opposite of somatic cell.
Germ plasm	-	The germinal material or physical basis of heredity.
Globulin	-	One of the common proteins in the blood, some of which like gamma globulins function as antibodies.
Gonad	-	A sexual gland - testis or ovary.
Guanine	-	A purine base found in DNA and RNA. Pairs normally with cytosine in DNA.
Gynandromorph	-	Gynander - a sex mosaic which is sterile. An individual having some cells with xx sex compliment and others with xy compliment.
Haemoglobulin	-	Oxygen - transporting protein present in red blood cells, composed of two pairs of iron containing identical polypeptide chains.
Haemophilia	-	Bleeders disease - A tendency to profuse bleeding even from a slight cut due to a recessive gene on the x chromosome.
Haploid	-	Monoploid - An individual or cell having a single set of chromosomes.
Hardy-Weinberg law	-	In a large random mating population, the gene and genotypic frequencies remain constant from generation to generation in the absence of migration, mutation and selection. Mathematical relationships between allele frequencies within the population are described by the expansion of the binomial equation $a^2+2ab+b^2$.
Helix	-	A structure with the spiral shape. The Watson and Crick model of DNA is double helix form.
Hemizygous	-	The condition in which only one allele of a pair is present as in sex linkage or as a result of deletion.
Heritability	-	The ratio of additive genetic variance to the total phenotypic variance. Symbolised as h^2.
Hermaphrodite	-	An individual with both male and female reproductive organs.
Heterochromatin	-	Inactive and condensed form of chromatin. Stains differently and functioning differently than the euchromatin that contains the genes. The nucleolar organizing region, the site of ribosomal RNA synthesis is flanked on either side by heterochromatin.
Heteroduplex	-	A DNA molecule in which the two strands do not have complementary base sequences.
Heterogametic sex	-	That sex having two different sex chromosomes as XO, XY or ZW. The heterogametic sex produces two kinds of gametes eg. x and y sperms in man.
Heteroploid	-	Aneuploid - An individual having a chromosome number other than the true haploid or diploid number (2n +1 or 2n-1). Opposite of Euploid.
Heteropyknosis	-	Property of certain chromosomes or of their parts to remain more dense and to stain more intensely than other chromosomes or parts during the nuclear cycle.

Heterosis	Hybrid vigour - Superiority of heterozygotes with respect to one or more traits in comparison with corresponding homozygotes.
Heterozygote	An individual with unlike members of any given pair of alleles. They produce more than one kind of gamete with respect to a particular locus.
Hexaploid	Chromosome number of 6n, having six sets of chromosomes.
Hfr	High frequency of recombination of E.coli strains. In these cells the F factor is integrated into bacterial chromosome.
Histones	A group of proteins rich in basic amino acids specially arginine and lysine. They function in the coiling of DNA in chromosomes and in regulation of gene activity.
Holandric gene	A gene located only on the Y chromosome in XY species and thus transmitted from father to son.
Homogametic sex	A sex producing similar gametes.
Homologous chromosomes	Chromosomes occurring in pairs, one derived from each of two parents, morphologically similar in size and shape and bearing same gene loci.
Homozygote	An individual whose chromosomes bears identical genes of a given allelic pair. Homozygotes produce only one kind of gamete with respect to particular locus and hence breed true.
Hybrid	An offspring of a cross between two homozygous parents, a heterozygous individual.

Hybridization	Crossing of two genetically different individuals.
Hypertrichosis	Long hair growth on ears - Y linked character.
Idiogram	A diagrammatic representation of the complete chromosome set of an individual arranged according to size and or accepted numbering system.
Immigration	Introduction into the population.
Immune reaction	The production of antibodies in response to antigens.
Immunoglobulins	Antibody molecules. There are five classes - IgG, IgM, IgA, IgD and IgE.
Inborn errors of metabolism	Inherited diseases that can be called as genetic blocks in specified metabolic pathways, usually due to recessive alleles determining a decreased activity or the absence of albinism or alkaptonuria.
Inbreeding	Matings among related individuals.
Inbreeding coefficient	The proportion by which inbreeding has reduced heterozygosity in a given population. The probability of a descendant being homozygous or two alleles at any locus being identical by descent; percentage of all loci which were heterozygous in the base population that now have become homozygous due to inbreeding.
Incompatability	The presence of antigens in a foetus or donor blood that can induce an immune response in the mother or the recipient.

Incomplete dominance	A type of inheritance where the heterozygous individual shows the characteristics midway between the homozygous parents.
Independent assortment	The random distribution of genes to the gametes that occurs when genes are located in different chromosomes.
Inducer	A molecule that binds to a repressor, preventing it from blocking transcription.
Induced mutation	A mutation due to a specific treatment.
Induction	Phenomenon of separation of temperate phage genetic material from E.coli chromosome.
Inhibitor	A major or modifier gene that interferes with a reaction.
Initiator codon	First, translated codon in mRNA 5'AUG 3'.
Intercross	A cross between two heterozygotes of the same alleles.
Interference	The phenomenon that when a crossing over occurs at one point of chromosome another crossing does not occur within a certain distance of it.
Interferon	Protein produced in an animal cell when infected by a virus and which inhibits viral multiplication.
Interphase	The stage of cell cycle during which cell is not dividing, the metabolic stage following telophase of one division and preceding the prophase of next division, G1, S, G2 phase of the cell cycle, resting stage between two mitotic divisions.

Intersex	An individual showing secondary sex characters intermediate between male and female.
Inversion	Reversal of a segment of a chromosome and the linear arrangement of genes within that segment lie in an inverse order.
Isoagglutinogen	An antigen like A or B blood type factor, that occurs normally.
Isochromosome	A chromosome with two morphologically identical arms.
Isozymes	Enzymes performing similar functions.
Kappa particles	Particles in the cytoplasm of paramecium that grow in the presence of gene K, producing a killer type.
Karyokinesis	The division of nucleus during cell division.
Karyotype	The chromosome constitution of a cell or an individual. Metaphase chromosomes arranged in order of length and position of centromere. Also the abbreviated formula for chromosome constitution.
Klinefeleter's syndrome	Abnormal phenotypic male due to the presence of one or more extra x chromosomes along with a y chromosome.
Lambrush chromosome	Large diplotene chromosomes having paired loops extending laterally present in primary oocyte nuclei of many animals representing active sites of RNA synthesis or transcription.
Leptotene	Early stage in meiotic prophase when the individual chromosomes appear as fine thread like structures.

Lethal	A genetic condition which causes the premature death of an organism.
Lethal gene	A gene which causes visible abnormality or drastic effect to kill the bearer. Death may occur at any time from fertilization to advanced age. It may be dominant, recessive or incompletely dominant.
Ligase	An enzyme capable of joining two segements of a broken strand of double-stranded DNA.
Line breeding	Mating of selected members of successive generations among themselves to fix desirable characteristics.
Linkage	The occurrence of different non-allelomorphic genes on the same chromosome. They are transmitted as one group and they fail to exhibit independent assortment.
Linkage map	Genes of a given species listed in linear order to show their relative positions on the chromosomes.
Locus	A specific position occupied by a gene or one of its alleles on a chromosome.
Lysis	Bursting of a cell by disintegration or dissolution of a cell membrane.
Macromolecule	A large molecule - the term used to identify molecules of proteins and nucleic acids.
Major gene	A gene that may cause sufficiently large variation in the trait, which can be easily detected.
Map unit	A distance on a linkage map represented by one per cent of crossing over.

Maternal effect	Trait influenced by a gene of the mother but expressed in the progeny.
Maternal inheritance	Inheritance of characters influenced by extra chromosomal i.e. cytoplasmic factors.
Maturation	The formation of gametes.
Meiosis	Nuclear division in which the diploid chromosome number of reproduction cell is reproduced by half. Results in the formation of gametes. Important source of variability through recombination.
Modifier	A gene that affects the expression of another gene. (modifying gene).
Monohybrid	An offspring of two homozygous parents that is heterozygote for a pair of alleles.
Monohybrid cross	A cross between two individuals differing in only one heritable trait.
Monoploid	Organism or cell having a single set of chromosomes (n).
Monosomic	An individual lacking one chromosome of a set (2n-1).
Monozygous twins-	Twins that develop from a single zygote. They have identical genotypes.
Morphology	Study of the form of an organism.
Mosaicism	The presence of two or more cell genotypes in an individual arising by mutation or by chromosomal aberration.
Mulatto	Offspring of a cross between pure negro and pure White parents, thus heterozygous for skin colour alleles.

Multiple alleles	Sets of alleles having three, four and even twenty alleles.
Multiple genes	Several sets of alleles producing more or less equal and cumulative effects on the same character.
Mutagen	A chemical or physical agent that induces mutation in a gene.
Mutation	A sudden, stable change in the gene structure responsible for change in character.
Mutation pressure	A constant mutation rate that adds mutant genes to a population.
Mutator	Allele or gene that causes other genes to mutate.
Muton	The smallest unit within a gene or a subunit of a cistron, alteration of which can cause a mutation.
N	Haploid genome.
Natural selection	Natural processes based on the principle of the survival of the fittest.
Non-disjunction	The failure of homologous chromosomes to separate at anaphase of meiosis or mitosis. One daughter nucleus receiving two chromosomes and other none.
Non-linked genes	Genes which are located on different chromosomes and segregate randomly.
Nonsense codon	A codon that does not designate any amino acid in the genetic code. A codon for termination of translation in a polypeptide chain.
Normal curve	A smooth, symmetrical bell shaped curve of distribution.

Nuclease	Enzyme that breaks down nucleic acids.
Nucleic acid	Polynucleotide: hereditary material. An acid having phosphoric acid, pentose sugar and organic bases. DNA and RNA are nucleic acids.
Nucleolar organizer	A chromosomal segment that controls the synthesis of ribosomal RNA.
Nucleolus	A spherical structure present in nucleus involved in rRNA synthesis. Associated with the nucleolar organizer.
Nucleoprotein	Compound of nucleic acid and protein. Material of which chromosomes are made.
Nucleoside	Component of nucleic acid made of one deoxyri- bose molecule in DNA or ribose in RNA plus purine or Pyrimidine.
Nucleotide	A building unit of nucleic acid containing a phosphate, a sugar and an organic base.
Nucleus	Part of a cell containing the chromosomes and surrounded by cytoplasm.
Nullisomic	An abnormal organism lacking both members of a chromosome pair (2n - 2).
Octoploid	An organism or cell with eight sets of chromosomes, a polyploid.
Oncogenic	Having a tendency to cause cancer.
Oocyte	Unfertilized egg cell, cell which undergoes two meiotic divisions to form ovum (oogenesis).
Oogenesis	The formation of ovum in animals.
Oogonium	A germ cell of the female before meiosis begins.

Operator gene	A DNA segment in an operon that regulates the transcription of one or more structural genes.
Operon	A genetic unit consisting of an operator site, a promotor and one or more structural genes whose activity is influenced by the operator.
Organelle	A subcellular membrane - bound structure with a particular function.
Organism	An individual characterized by its interdependent genetic material and proteins.
Outbreeding	Mating of unrelated or less related individuals.
Over dominance	A condition in which the heterozygote exhibits a more extreme manifestation of the trait than does either homozygote.
P	Symbolizes the parental generation.
Pachytene	A mid prophase stage in meiosis immediately following zygotene and preceding diplotene.
Panmixia	Random mating in a population.
Paracentric inversion	An inversion in which both breaks occur on the same arm of the chromosome and that does not include the centromere.
Parameter	A value based on an entire population.
Parthenogenesis	Development of a new individual from an unfertilised egg.
Paternal	Pertaining to the father.
Pedigree	The ancestral history of an individual.
Penetrance	The degree to which a specified genotype expresses a phenotypic effect.

Peptide	A string of few amino acids joined by peptide bonds.
Peptide bond	A chemical bond (CONH) linking amino acid residues together in a protein.
Pericentric inversion	An inversion including the centromere, breaks occurring on the opposite sides of centromere.
Phage	A virus infecting a bacterial cell, bacteriophage.
Pharmacogenetics	The study of relation between individuals genotype and its reaction to various pharmaceutical agents.
Phenocopy	An individual who has a phenotype similar to that produced by a certain mutant genotype even though the individual may not have the genotype.
Phenotype	The outward, visible expression of the hereditary constitution of an organism. It is usually expressed in words eg. Red, Roan, White etc.
Phenylketonuria	Metabolic disorder resulting in mental retardation, transmitted as a Mendelian recessive.
Phylogeny	The evolutionary development of a species.
Plasmagenes	Self-replicating cytoplasmic particle's units believed to be responsible for some extranuclear inheritance.
Plasmid	Dispensible chromosome existing and replicating autonomously in the cytoplasm of cells, either artificially or naturally but can not be integrated into a chromosome of the host.
Plastids	A cytoplasmic body present in the cells of plants and some protozoa.

Pleiotropy	The influencing of more than one trait by a single gene.
Ploidy	Variation in the number of chromosomes.
Polar bodies	In females, the smaller cells produced at meiosis that do not develop into ovum.
Polydactyly	The occurrence of more than the usual number of fingers or toes.
Polygene	One of a series of multiple genes involved in Quantitative inheritance.
Polymer	A compound composed of many smaller subunits.
Polymerase	An enzyme that catalyzes the formation of DNA or RNA.
Polymerization	Chemical union of two or more molecules of the same kind.
Polymorphism	The occurrence of two or more alleles for a given locus in a population.
Polynucleotide	A linear sequence of joined nucleotides in DNA or RNA.
Polypeptide	A compound containing two or more amino acids joined by peptide bonds. A protein may have one or more specific polypeptide chains.
Polyploid	Organisms which have more than two full sets of homologous chromosomes (Triploid 3n, tetraploid 4n, pentaploid 5n, hexaploid 6n, hepta ploid 7n, Octoploid 8n).
Polyribosomes	A group of ribosomes linked together in a chain by a single long molecule of messenger RNA.

Polytene Chromosomes	Giant chromosome produced by interphase chromosomes replication without division and consisting of many identical chromatids.
Population	The group of individuals of a given species inhabiting a specified geographic area.
Population (Effective)	Breeding members of the population.
Population genetics	The branch of genetics that deals with frequencies of alleles and genotypes in breeding populations.
Position effect	A change in the phenotypic expression of a given gene as a result of change of its position in the chromosome or with respect to neighbouring genes.
Preformation	The hypothesis putforth by Jan Swammerdam who believed that the egg unfolds the characters already present in it in a miniature form.
Primer	Nucleic acid synthesis starting point, the 3' end of a single stranded piece of nucleic acid.
Progeny	Offspring individuals.
Prokaryote	Simple unicellular organism lacking nuclear membrane and having chromosomal material in the form of haploid DNA without the important constituent of protein, as in eukaryote.
Promoter	Part of DNA at which RNA polymerase binds transcriptase and initiates transcription.
Prophage	A non-virulent bacteriophage DNA integrated into the DNA of the chromosome of host bacteria.
Prophase	The first stage of nuclear division including events from the first appearance of the chro-

	mosomes to their arrangement at the equator of the spindle.
Protein	A very large complex organic molecule having a single polypeptide chain.
Proteolysis	The breakdown of proteins.
Pseudoalleles	Non-alleles so closely linked as often to be inherited as one gene.
Pseudo dominance	The expression of a recessive gene at a locus.
Pure line	A population obtained by continuous inbreeding over many generations, a line in which homozygous individuals produce only homozygous offsprings like parents.
Purine	A double ring type nitrogenous base component of DNA and RNA (Adenine and Guanine).
Pyrimidine	Single ring type nitrogenous base found in DNA (thymine and cytosine) or RNA (uracil and cytosine).
Qualitative trait	A trait which differs sharply among individuals and can be easily classified into discrete classes. Depends on one or few sets of alleles.
Quantitative trait	A trait that varies continuously and depends on cumulative action of many genes.
Races	Subdivisions of a species, recognizably different from one another.
Radioactive isotope	An unstable isotope (form of an atom) that emits ionizing radiation.
Random drift	Variation in gene frequency due to chance fluctuation.

Random mating	Each individual has an equal chance of mating with other individual within the population with no selection in operation.
Random sample	A sample obtained in such a way that each individual in the population being studied has the same chance of being selected .
Recessive genes	A gene exhibiting no expressivity in the phenotype in the presence of its dominant allele.
Reciprocal cross	A second cross of the same genotypes in which the sexes are reversed. For eg. a female from strain Ax a male from strain B and a male from Ax a female from B are reciprocal crosses.
Reciprocal translocation	The exchange of segments between two non-homologous chromosomes or of unequal segments between homologous.
Recombinant	An individual possessing a combination of traits from both parents. An individual obtained from a cross over gamete.
Recombination	A new linkage other than the parental ones formed by crossing over.
Recon	The smallest segment of DNA or subunit of a cistron that is capable of recombination.
Recurrent mutation	The mutation occurring frequently.
Reduction division	Meiotic division in which the maternal and paternal elements of the bivalent separate.
Regression	The correlation between parents and offspring or other related individuals.
Regulator gene	A gene that controls the rate of synthesis of the product of the other gene. A gene responsible for 'switching on or off' other genes.

Sense codon	Codon specifying an amino acid in protein synthesis.
Sense strand	DNA strand employed as a template for transcription.
Sex chromatin	Barr body. A densely staining body visualised in cells with more than one x chromosome. An inactivated x chromosome of normal mammalian females but not males.
Sex chromosomes	Chromosomes not occurring in identical pairs in both sexes. They determine the sex of the individual and are designated x and y chromosomes in man, animals, and Drosophila and as z and w in birds.
Sex duction	The incorporation of bacterial genes by sex factors and their subsequent transfer by conjugation to a recipient cell.
Sex influenced trait	One in which dominance of an allele depends upon the sex.
Sex limited trait	The trait which is expressed in only one of the sexes.
Sex linkage	Association of a hereditary trait with sex.
Sex linked gene	A gene located only on the x chromosome in xy species or on the z chromosome in zw species.
Sexual reproduction	Reproduction involving the formation of mature germ cells - eggs and sperms.
Sibs	Individuals having the same maternal and paternal parents. Brother-sister relationship.
Sib mating	Brother-sister matings.

Sickle cell anaemia	A severe genetic disorder due to an abnormal haemoglobin causing a marked change in the shape of RBCs to sickle shaped. A recessive lethal allele produces a mild anaemia in heterozygous condition, and in homozygous condition, it may cause lethal anaemia.
Sister chromatids	Chromatids of the same chromosome.
Somatic	Non-reproductive tissue or cell or a protoplasm, in contrast to germplasm.
Somatic cells	Referring to body tissues, having two sets of chromosomes.
Species	A set of individuals who can interbred and produce fertile progeny.
Sperm	A mature male germ cell.
Spermatid	The four cells formed by the meiotic divisions in spermatogenesis.
Spermatocyte	The cell that undergoes two meiotic divisions to form four spermatids.
Spermatogenesis	The process by which maturation of the gametes (sperm) of the male takes place.
Spermatogonium	Primordial male germ cell that may divide by mitosis to produce more spermatogonia.
Spermiogenesis	Formation of sperm from spermatids.
Spindle	A bipolar molecularly oriented structure, composed of microtubules made of protein subunits, taking part in chromosome movement at nuclear divisions during mitosis and meiosis.

Standard deviation	-	A measure of variability in a population of individuals.
Standard error	-	A measure of variation of a population of means.
Sterility	-	Inability to produce offspring.
Structural gene	-	A cistron. A gene that controls actual protein production by determining the amino acid sequence.
Sublethal gene	-	A lethal gene with the delayed effect.
Synapsis	-	The pairing of homologous chromosome occurring in prophase I of meiosis.
Synaptinemal complex	-	A ribbon like structure formed between synapsed homologous at the end of prophase I of meiosis, binding the chromatids along their length and facilitating chromatid exchange.
Syndrome	-	A group of symptoms that occur together and represent a particular disease.
Synteny	-	The occurrence of two loci on the same chromosome.
Telomere	-	Terminal gene which prevents a chromosome being joined to any other chromosome end.
Telophane	-	A chromosome with the centromere located at one end.
Telophase	-	The last of the four stages of mitosis during which the daughter nuclei appear and the cytoplasm usually divides.
Template	-	A macromolecule that serves as a mould and provides the information for the synthesis of a complement.

Termination Factor (TF)	-	Protein required to obtain release of newly synthesised polypeptide chain from tRNA.
Terminalization	-	Repelling movement of the centromeres of bivalents in the diplotene stages of the meiotic prophase that tends to move the viable chiasmata towards the ends of bivalents.
Test cross	-	Back cross to the recessive parental type or a cross between genetically unknown individual and a homozygous recessive individual used for determining heterozygosity. Or as a test for linkage.
Tetrad	-	A bundle of four homologous chromosomes produced at the end of the first meiotic prophase.
Tetraploid	-	Cell or individual with four haploid sets (4n) of chromosomes.
Tetrasomic	-	A nucleus or an organism with four members of one of its chromosomes whereas the remainder of its chromosome complement is diploid (2n+2).
Thymine	-	A pyrimidine base found in DNA. Pairs normally with adenine.
Totipotency	-	An undifferentiated cell such as blastomere that when isolated develops into a complete embryo.
Trait	-	Character or form .
Trans-arrangement	-	Linkage of the dominant allele of one pair and the recessive allele of another on the same chromosome. Also known as repulsion.
Transcriptase	-	DNA-dependent RNA polymerase.

Transcription	Synthesis of messenger RNA using a DNA template.
Transduction	Transfer of bacterial genes from one bacterium to another through the agency of bacteriophage genetic recombination whereby DNA is transferred from one cell to another through the mediation of a plasmid.
Transferrin	Blood serum protein, betaglobulin.
Transfer RNA (tRNA)	RNA that transports amino acids to the ribosome where they are assembled into proteins also called soluble or adapter RNA.
Transformation	The genetic recombination whereby naked DNA from one cell or virus becomes incorporated and integrated into another cell or virus.
Transgressive variation	Appearance in progeny of a more extreme expression of a trait than that occurs in the parents. Assumed to result from cumulative action of multiple genes.
Transition	A mutation caused by the substitution of one purine by another purine or one pyrimidine by another pyrimidine in DNA or RNA.
Translation	Formation of a chain of amino acids during protein synthesis as a result of information and directions contained in mRNA.
Translocation	The shift of a segment of a chromosome to another part of the same chromosome or to a different chromosome.
Transversion	A mutation as a result of the substitution in DNA or RNA of a purine for a pyrimidine or vice versa.

Trihybrid	An individual heterozygous for three pairs of genes.
Triploid	An individual having three sets of chromosomes (3n).
Trisomic	A cell or organism with an extra somatic chromosome over the diploid-number (2n+1).
Trivalent	Three synapsed homologous chromosomes.
True breeding	Producing a progeny having exactly the same genotype as the parents.
Turner's syndrome	A human abnormality in an individual who is phenotypically female, but characterised by the absence or non-functional ovary and xo chromosome constitution.
Univalent	A chromosome unpaired at meiosis.
Unreduced gamete	A gamete having the somatic (2n) chromosome number instead of half that number (n).
Uracil	A pyrimidine base occurring in RNA but not in DNA.
Value	The observed character of the individual.
Variance	The square of the standard deviation. A measure of variation.
Variation	The occurrence of differences among the individuals of the same species.
Viability	Degree of capability to live and develop normally.
Virulent phage	A phage (virus) that destroys the host (bacterial) cell.
W chromosome	One of the sex chromosomes of heterogametic female.

Wild type	The phenotype most frequently found in nature, the normal phenotype.
Wobble hypothesis	Phenomenon that explains how the anticodon of one tRNA may recognise two or more codons.
X chromosome	A chromosome associated with sex determination. In animals, Drosophila and human beings, the female has two and the male has one x chromosome.
X inactivation	The genetic inactivation of all x chromosomes in excess of one.
X linked gene	A gene located on the x chromosome. A trait determined by such a gene is called x linked trait.
Y chromosome	The partner of the x chromosome in the male of many species. In Drosophila it is composed mostly of heterochromatin and carries few genes, but does not influence maleness. In human, it carries the genes which influences maleness.
Y-linked gene	A gene located on the y chromosome. A trait determined by such a gene is called a y linked trait.
Z chromosome	Used for y chromosome where the female is heterogametic.
Zygote	The protoplast formed by the union of gametes. A fertilized egg.
Zygotene	The stage in meiosis during which synapsis occurs. After the leptotene and before the pachytene stage in meiosis.

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